

**YOUNG PEOPLE'S EXPERIENCES OF  
NEUROFIBROMATOSIS TYPE 1**

**JENNY BARKE**

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## Abstract

Neurofibromatosis type 1 (NF1) is a genetic condition which can result in varying degrees of visible difference (disfigurement). There is currently very little research into the psychosocial impact of NF1, particularly during adolescence, a time when health behaviours are consolidated and appearance concerns become more salient. While clinical reviews, research and case studies have suggested that appearance is likely to play an important role in the lives of people with NF1, how young people manage appearance concerns and the possibility of a changing appearance on a day-to-day basis has not specifically been researched. The impact of appearance changes (and potential changes) associated with the condition are therefore an important area to understand further.

In order to explore the role of appearance within young people's experiences of NF1 this thesis takes an exploratory qualitatively driven mixed methods approach. In-depth interviews were carried out with 19 people; nine young people aged 14-24 with a diagnosis of NF1; seven parents of young people with NF1 and three health professionals who work with people with NF1. Interviews were thematically analysed and subsequent themes informed the development of mixed methods surveys of young people with NF1 (n=73), parents (n=55) and health professionals (n=53).

Both interview and questionnaire data highlighted the variety in young people's experiences of NF1. While appearance was important to young people, this was primarily in terms of general appearance evaluations and managing the uncertainty of possible future changes to their appearance rather than how noticeable they felt the condition was. While health professionals agreed with this assessment, an important difference between participant groups was that parents felt that the noticeability of symptoms impacted on their child's experience. All participant groups underlined the importance of managing social interactions and the reactions of other people. Participants also highlighted a need for greater awareness and understanding of NF1, and access to trustworthy information and advice about their condition.

Suggested applications from findings discussed in this thesis include providing young people with age appropriate information regarding their condition, in different formats including access to health professions who are specialists in genetics or NF1. Young people may also benefit from targeted help and support related to social skills, specifically including support in talking about NF1 and answering questions about their condition. It is suggested that future research should explore how young people with NF1 and their parents manage uncertainty and noticeability.

This thesis contributes to the literature by providing data from groups who are under researched about an aspect of their experience that has not been explored. By employing a range of methods and including different participant groups an in-depth, detailed understanding of the role that appearance plays within young people's experiences of NF1 is provided.

## **Approach of this thesis and chapter outlines**

This thesis aims to provide an in depth exploration of the role that appearance plays within young people's day-to-day lived experiences of the genetic condition Neurofibromatosis type 1 (NF1). It investigates the challenges facing young people with NF1, with regards to the role of appearance, and how they manage these challenges. This is explored from the perspective of young people themselves, their parents and health professionals working in this field. Before reviewing the relevant literature and considering methods of investigation this brief section provides a personal account of my approach to this research.

Mauthner and Doucet (2003) highlight the importance of recognising that as researchers we have our own academic and personal biographies that shape our research. Personal experience of a topic can be used as a way to engage enthusiastically with an area of research (Finlay, 2002). As such, in terms of my own experience it is important to acknowledge that I have personal experience of being a young person with a chronic health condition and of parenting a child with a chronic health condition. I was diagnosed with Juvenile Idiopathic Arthritis (JIA) when I was three years old, this mainly affected my legs and I had swollen knee joints fairly often. Looking back I do not remember ever being concerned about my JIA. I was never very sporty and loved the fact that JIA was a ready-made excuse to miss PE and sports days; if I said it hurt I could miss any sports I did not enjoy (namely rounders, hockey and cross country) although I was fine to take part in ice-skating and skiing. I also enjoyed having time off of school for hospital appointments and was lucky to have the same health care team through childhood and adolescence. My transition to adult care was not quite so simple, I stayed as a paediatric patient until my early 20's and then did not see any professionals for several years.

However, overall my experience of growing up with JIA was fairly positive. Another area of my life in which I have personal experience of managing chronic health conditions is as a parent. I have two children, a daughter and a son. When my son was a very young baby he was diagnosed with renal failure and whilst in hospital a hemi vertebra was found (and a second later). He has chronic renal failure and congenital scoliosis; he also had some necrosis to his hip. As a parent, knowing your child has any kind of medical issue is always a huge concern, both in terms of managing the medical situation and also wanting to make sure you and your child and your family (including other children) manage emotionally and remain positive and resilient. I feel my own personal experience, both of having an ongoing health condition through adolescence and being a parent of a child with a chronic health condition, provide me with a personal insight into some of the areas I am researching. Experience differs and I would not presume that my own experience, as an individual or as a parent is the same as other families and individuals. However, I feel that it is important to acknowledge that my own personal experience is likely to inform the way in which I approach and engage in this research.

My professional experience is also highly relevant to this research. Prior to starting this PhD, most of my work has been with young people and families. While the work has been in different environments and contexts a clear thread running through my work has been the importance of engaging with and

empowering young people and their families. I am very passionate about the importance of understanding and listening to young people and this is likely to influence and guide the approach I take to this research.

In addition to my own experience, it is important to note that this research was undertaken at the Centre for Appearance Research (CAR), a research centre at the University of the West of England in Bristol. Research at CAR involves psychological and interdisciplinary research in appearance, disfigurement, body image and related studies. CAR aims to “*make a real difference to the lives of the many hundreds of thousands of people with appearance-related concerns both in the United Kingdom and across the world*” (CAR, 2013). The centre aims to achieve this through increasing knowledge, understanding and awareness of the psychosocial aspects related to appearance and visible difference.

## **Language**

Appearance is potentially a sensitive area to research and young people are often seen as a vulnerable group. As such, much attention has been paid to the use of language in this thesis. Terminology used in ‘disfigurement’ research can be negative and often has biomedical connotations (e.g. deformed, abnormality). This thesis recognises that in some forms of dissemination it may be that the term ‘disfigured’ is used as it is clearly understood, for instance in medical contexts. However this will be used in order to then move on and promote language that is more positive such as ‘visibly different’ and ‘altered appearance’. This borrows from previous research at the Centre for Appearance Research including ‘Guidelines for Appearance Research’ (CAR, 2013) and the preface in ‘The Psychology of Appearance’ (Rumsey & Harcourt, 2005).

## **Thesis Structure and Chapter outlines**

This research takes a qualitatively driven mixed methods approach to researching the role appearance plays within young people’s experiences of Neurofibromatosis type 1 (NF1). The starting point of this thesis was talking to young people, their parents and health professionals with the aim of gaining an understanding of the issues that were important within young people’s experiences of the appearance-related aspects of NF1 from their own perspectives, as well as parents and health professionals. This knowledge was used in order to develop focused questionnaires that explored specific areas identified in the interview studies in a survey.

This thesis is comprised of four sections. The first section (Chapters 1, 2 & 3) situates the research, reviews current literature related to young people and chronic illness, appearance and the psychosocial impact of NF1 and then outlines the methods used. The following section (Chapters 4, 5 & 6) details interview studies with young people, parents and health professionals. The third section (Chapters 7 & 8) describes how findings from interview studies were used to develop questionnaires

which surveyed young people, parents and health professionals. The final section (Chapters 9) discusses the thesis findings and evaluates the implications of these findings and the methods used.

The chapters are outlined in greater detail below:

## **Section 1: Situating the research**

**Chapter 1** presents an overview of Neurofibromatosis type 1 (NF1) and considers how this thesis defines 'young people' before considering the factors that impact on the management of chronic health conditions during this developmental stage.

**Chapter 2** considers the psychosocial impact of NF1 across the lifespan and explores why appearance is important to people. Literature relating to adjustment to a visible difference and young people's experiences of visible difference is reviewed leading to a consideration of why an understanding of the role of appearance within young people's experiences of NF1 is needed.

**Chapter 3** considers how to research young people's experiences. The aims and objectives of this research are outlined and discussed, following which the methods used across the thesis are described. The rationale for adopting a mixed methods approach is discussed and the ethical and practical implications of conducting research with young people are considered.

## **Section 2: Interview studies**

**Chapters 4, 5 and 6** detail exploratory in-depth interview studies. Chapter 4 considers the perspectives of young people with NF1 themselves, Chapter 5 details interviews with parents and Chapter 6 is an account of interviews with health professionals. Each chapter presents the aims, method and findings of the individual study.

## **Section 3: Survey development and findings**

**Chapter 7** details the development of three surveys for (1) young people, (2) parents and (3) health professionals. The chapter presents the aims of this research phase and the research questions. Following this, how findings from the interview studies have informed the development of a questionnaire for each participant group is considered. Instruments used to measure aspects identified as warranting further investigation are introduced and the format of each questionnaire is outlined and discussed.

**Chapter 8** describes the ethical approvals, recruitment details and procedure of the survey study. The study findings are presented in four sections; (1) the young people's survey (2) parents' survey, (3) comparisons between young people and parent measures and then the health professionals' survey (4).

## **Section 4: Discussion**

**Chapter 9** discusses the findings of this thesis overall. The role that appearance plays within young people's experience of NF1 is discussed and the chapter then considers the implications of the thesis findings in terms of care and support for young people with NF1. The methods used in this thesis are evaluated and recommendations for further research are outlined. Finally the contribution of this thesis is summarised.

## Chapter 1: Introduction

This introduction serves to outline and define the main topics covered in this thesis. An overview of Neurofibromatosis type 1 (NF1) is presented, followed by a consideration of what is meant by 'young people' and the factors that impact on the management of chronic health conditions during this developmental stage.

### 1.1 What is Neurofibromatosis Type 1?

This section describes the aetiology, symptoms, history, diagnosis and management of Neurofibromatosis Type 1 (NF1). The psychosocial impact of the condition is outlined and discussed in Chapter 2.

Neurofibromatosis Type 1 (NF1) is one of the most common genetic disorders (Riccardi & Smirniotopoulos, 1992) affecting 1 in 2500 to 1 in 3000 individuals (Ferner et al, 2012). It is as prevalent as Down's syndrome and cystic fibrosis and twice as common as muscular dystrophy (Counterman et al, 1995). Approximately 50% of cases are inherited from a parent and 50% are de novo (new mutations). Individuals with NF1 have a 50% chance of their child inheriting the condition (Ferner et al, 2007).

NF1 is caused by a genetic mutation which can cause tumors to grow on the nerves; these cutaneous neurofibromas are benign tumors which grow on the skin, often first appearing during the hormonal changes that occur in teenage years. They vary in size and can appear anywhere on the body. The number of neurofibromas present differs greatly between individuals and over a lifetime (Duong, 2011). Café au lait spots are often the first and most common sign of NF1, these are often found at birth and are usually evident by two years of age. Ten percent of the population have these marks on their skin (Ferner et al, 2007), however nearly every person with NF1 has six or more of them. The number of Café au lait spots may increase in childhood and occasionally later in life. Other features of NF1 can include Lisch nodules of the iris (small pigmentation in the iris which causes no disturbance to vision), skin-fold freckling (freckling/pigmentation in the groin and armpits), plexiform neurofibromas (or sub cutaneous neurofibromas) which are diffuse tumors that grow along a nerve and are found in at least 25% of people with NF1 (Huson, 1989), optic gliomas (tumor of the optic nerve) and skeletal complications including pseudoarthrosis (meaning false joint) and scoliosis (curvature of the spine). Macrocephaly (large head size) is also common and short stature is found in around a third of people with NF1 (Hersh, 2008). Rare complications include a risk of malignancy, organs being compromised by neurofibromas, seizures and hypertension (Ferner, 2012). In most instances NF1 is obvious by the age of one and penetrance is 97% by eight years (DeBella et al, 2000). Specific problems usually develop by 20 years. As a rough guide, Riccardi and Smirniotopoulos (1992) suggests 33% of people with NF1 are minimally affected, 33% have mild to severe problems and 33% are severely affected. Cognitive deficits are the most common complication in children with NF1 (Hyman et al, 2006). Learning difficulties, Autistic Spectrum Disorders, Attention Deficit Hyperactivity Disorders and sleep and behavioural difficulties occur more frequently in children with NF1 than in the general population

(Garg et al, 2013; Johnson et al, 2005; North et al, 1997). It is estimated that between 30-65% of children with NF1 may have learning difficulties (Cutting et al, 2004).

Genetic testing for NF1 is possible, but it is not generally recommended, because there is little link between the genotype and phenotype, meaning that genetic testing tells an individual very little about how they may be affected (Boyd et al, 2009). Symptoms vary greatly and severity cannot be predicted, even within families (Cnossen et al, 1997). Therefore, diagnosis is generally based on the NIH guidelines (see figure 1) which were established at the 1987 international consensus conference and reaffirmed after review in 1997.

**The diagnostic criteria for NF1 are met in an individual if two or more of the following are found:**

- Six or more cafe-au-lait macules on the skin
- First degree relative with NF1
- Two or more neurofibromas or 1 plexiform neurofibroma
- Anxillary/groin freckling
- Two or more lisch nodules (in the iris)
- Bony dysplasia (abnormal bone growth)
- Optic pathway glioma (a type of brain tumor)

Figure 1: NIH Diagnostic criteria for NF1 (NIH Consensus Development Conference 1987 (cited in Huson, 1989))

Images of people with NF1 are thought to have been around since the 14<sup>th</sup> century. These early images depicted individuals as 'monsters'; they were frequently shown as distorted with possible symptoms of NF1 alongside mythological characteristics such as horns or talons (Brosius, 2010). The 1700's saw more scientific descriptions of NF1 and, in 1811, Louis Odier used the term neuroma to describe tumors on nerves (Brosius, 2010). The hereditary nature of the condition was highlighted by Virchow in 1847 (Rubenstein & Korf, 1990) whose student Von Recklinghausen recognised cafe au lait marks and freckling as symptoms of a larger disorder linked to tumors. The condition was then known as Von Recklinghausen's neurofibromatosis for many years. In 1900 Thomson assembled reports of instances within families (Riccardi & Smirniotopoulos, 1992) and surveys of the condition were published in the mid 20<sup>th</sup> century. In 1978 the National Neurofibromatosis Foundation was established in the USA. In the same year programs looking at NF1 as a distinct disorder were established and the disorder was classified into categories, not all have been accepted but NF1 remains as originally classified. NF1 diagnostic criteria and management guidelines were established in the United States by a National Institute of Health Consensus Conference in 1987.

An important part of the history of NF1 for many people is the link with Joseph Merrick, labelled "the elephant man", who for much of the 20<sup>th</sup> century was thought to have had the condition. This was first proposed by Dr Parkes-Weber in 1909, 19 years after Joseph Merrick's death. However, although confusion still exists (Legendre et al, 2011) researchers now believe Joseph Merrick may have had

proteus syndrome (a rare condition which involves, amongst other symptoms, atypical bone and skin growth). This link has been seen by some as useful and helpful in gaining publicity for the condition whilst others consider the persistent beliefs about the link between NF1 and 'the elephant man disease' to have had a negative impact on individuals' understanding and knowledge of the condition (Legendre et al, 2011).

There is still no cure for NF1. Treatment focuses on amelioration of symptoms and Rubenstein and Korf (1990) suggest there is no single approach to best care for people with the condition. The main aims of treatment are to support adjustment and to offer the best available medical treatment for specific complications. In terms of surgically managing symptoms, it is possible to surgically remove neurofibromas, however there is no guarantee that they will not grow back. Plexiform neurofibromas can also be managed surgically however nerve damage can occur and it is generally not possible to completely remove them as they can extend into tissue and bone. This means they can grow back after surgery (NHS, 2013). Other symptoms, including scoliosis and pseudoarthrosis can also be managed surgically or with orthopaedic interventions such as braces (NHS, 2013). It is important to note that these interventions may themselves bring challenges such as scarring and are unlikely to completely ameliorate symptoms.

The NIH Consensus Statement (cited in Huson, 1989) recommends annual examinations throughout life and suggests that parents and children need accurate information, psychological support and interactions with other families with the condition, underlining the need for clinicians to view involvement of the family as an essential aspect of care. Within the UK a consensus statement published by members of the UK Neurofibromatosis Association (now known as The Neuro Foundation) Clinical Advisory Board (Ferner et al, 2007) recommends that if a diagnosis of NF1 is being considered then a referral should be made to a clinician with knowledge of NF1 (geneticist, paediatrician, neurologist or dermatologist). Management in the UK consists of age-related monitoring, patient education and annual assessment of children in line with the NIH consensus statement. It is advised that young adults (16-25 years) receive education about NF1 and psychological support as neurofibromas often start to develop in late adolescence (Ferner et al, 2007). After mid 20's, monitoring depends on the individual and on the severity of the condition. Some continue to attend a specialist clinic, but the minimal requirement is for an annual blood pressure examination and patients should be aware of symptoms that need to be addressed should they arise. Similar advice is given in other international papers (including Hersh, 2008; Korf, 2001; North et al, 1997) all of which emphasise the importance of addressing the psychosocial needs of patients and recommend reviewing social adjustment and offering psychological support in addition to management of the physical symptoms of the condition.

In a handbook for patients, families and healthcare professionals, Rubenstein and Korf (1990) write that "*coping with neurofibromatosis is a challenge for both affected individuals and health-care professionals alike.....this begins with the medical burdens of the disorder and uncertainty about the future medical problems. Many affected individuals and their family members also face burdens of stigmatization, guilt and financial hardship that accompany a genetically determined variable medical*

*condition*" (xiii Rubenstein & Korf 1990). The psychological burden of NF1 is also stressed by Riccardi and Smirniotopoulos (1992) who suggest "*the vast majority of patients with NF1 beyond early and middle childhood suffer some compromise of their overall psychosocial performance as a direct and/or indirect result of their disease*" (1992: p195).

In terms of specialist care in the UK, The Neuro Foundation (a support group whose vision is to improve the lives of those affected by neurofibromatosis) part funds, along with the NHS, a small network of specialist advisors who provide support and advice for patients and families as well as a support and information line. Since 2009 there have been two nationally funded NHS complex care NF1 centres, at Guys and St Thomas' NHS Trust in London and at Central Manchester University Hospitals NHS Foundation Trust, which deliver coordinated care from specialist multidisciplinary teams.

Some people with NF1 may be offered genetic counselling with the aim of helping individuals "*understand and adapt to the medical, psychological and familial implications of genetic contributions to disease*" (Resta et al, 2006, p77). This often occurs at initial diagnosis in order to discuss the genetic nature of the condition with parents or may be offered later when people with NF1 start to consider having children of their own. Bernhardt and Halperin (in Rubenstein & Korf, 1990) suggest genetic counselling is the appropriate forum in which to address family education regarding medical genetics and family planning and to explore the psychological and emotional concerns that could arise when an individual or family member is diagnosed with NF1.

This section has described the condition NF1 and has outlined its management and treatment in the UK. The literature regarding the psychosocial impact of NF1 is considered in detail in Chapter two. The following sections outline how this thesis interprets the term 'young people' and then reviews current literature relating to young people's experiences of health.

## 1.2 Who are young people?

As this thesis explores the perspectives of young people it is useful at this point to define what is meant by the term 'young people' and explain why it is essential that their experiences and needs are considered. There are 1.2 billion people aged between 10 and 24 years of age; this equates to 1 in 5 people (WHO, 2013) and the health of this age group has improved much less than that of younger children in the last 50 years (Sawyer et al, 2012). Defining what is meant by 'adult', 'child' and 'young person' is complex. The UN convention on the rights of the child (2005) defines a child as a person under 18 years (suggesting therefore an adult is a person aged 18 or over). However, moving from childhood into adulthood is generally regarded as a process or a transition (Hogan & Astone, 1986). Whilst it is accepted that adolescence is a stage in-between or spanning childhood and adulthood, there is no definitive definition of the ages adolescence refers to. The World Health Organization defines adolescence as the life stage between the ages of 10 to 19 years (WHO, 2013) while the International Association for Adolescent Health defines adolescence as between the ages of 10 and 24 (IAAH constitution, 2013).

Entering adolescence is often associated with pubertal physical bodily changes; the term adolescence being derived from the Latin '*adolescere*', meaning to grow up. During adolescence individuals develop skills that will support them throughout adulthood. It is a life stage characterised by biological, cognitive and social change and is often portrayed as a difficult and challenging time. However, adolescence may be no more or less stressful than any other life period; in fact many young people are relaxed and happy through these years (Magen, 1998). While the physical changes of puberty are often seen as the start of adolescence, social transitions indicate the end of this period and entry into adulthood (Sawyer et al, 2012). The transition from adolescence into adulthood is generally associated with legal rights and social responsibilities. Becoming an adult is a subjective experience and is connected to individual qualities of character (Arnett, 1998). The transition to adulthood is marked by accepting responsibility for one's self, making independent decisions and becoming financially independent (Holmbeck, 2002; Arnett, 1997; 1998; Greene et al., 1992; Hogan & Astone, 1986). Becoming an adult may be occurring later in life than it has in the past and, in recognition of this, 'emerging adulthood' (Arnett, 2000; 2007) has been proposed as a distinct life stage to define those aged 18-24. This is a time, Arnett proposes, when people often do not see themselves as either children or as adults. However, there is some disagreement over whether the period from late teenage years into mid 20's is a distinct stage (Arnett, 2000; 2007) or whether it is young adulthood or extended youth (Bynner, 2005). While this stage can be referred to in many ways, including adolescence, emerging adulthood, youth, and young or early adulthood, it is useful to recognise that there is a stage between early teens and early/mid 20's that is somehow distinct to being a child or an adult. It is a time during which there is a great deal of change and development in an individual's life. During the early part of this stage people transition into adolescence and in the later part settle into adulthood. The importance of recognising the stages within development across adolescence and early adulthood are emphasised by Sawyer et al (2012), who note that, increasingly, the data of those in the age range of 10—24 year olds is divided into the three categories of early adolescence, late

adolescence and young adulthood. This division supports the application of research in young people's health; dividing this wide and diverse stage into subcategories may support more targeted research, interventions and care.

This thesis concentrates on the experiences of people aged 14-24 in order to explore mid-to-late adolescence and early adulthood. It is during this time that many of the physical characteristics of NF1 can change, individuals move from paediatric to adult health care, appearance becomes increasingly important to many individuals and furthermore, it is a time of great social and emotional change as people transition into adulthood. The term 'young people' is used in this thesis to refer to this age group whilst the terms 'adolescence' and 'early adulthood' are used to refer to the life stage.

### **1.3 Young people and chronic health conditions**

Having defined 'young people' for the purpose of this thesis, this section considers the importance of attending to their experiences of health. As outlined in section 1.1, NF1 is a lifelong chronic condition meaning that it is a long term illness that cannot be cured but rather one that individuals manage throughout their lives. This section seeks to outline the significance of adolescence and early adulthood in positive adaptation to chronic health conditions.

As young people transition from adolescence into adulthood, they generally begin to take more responsibility for their own healthcare. It is a time during which young people may become more aware of and learn more about their condition. It is a time when, in practical terms, young people may start to see clinicians on their own, rather than with their parents, and may start making their own decisions regarding treatment and management of their condition. These changes do not occur in a vacuum but are one part of a young person's life. This period of life is critical for establishing lifelong positive or risky health related behaviours, for all young people including those with a chronic health condition (Holmbeck, 2002). It is a time when positive health behaviours, such as exercise and diet, are established and health risk behaviours, such as smoking and drinking alcohol, may first be evident (Williams et al, 2002). However young people report feeling that they are not well served by health services (Jones & Bradley 2007) and the management of a chronic condition during adolescence is reported as challenging for young people, their families and health teams (Suris et al, 2004). Twelve percent of adolescents live with a chronic health condition, and this number is increasing (Sawyer et al, 2007). With advances in medical care the survival of children with chronic conditions into adulthood is becoming more common (Blum et al, 1993). Managing a chronic illness can be at odds with normative development during adolescence, however this is a time when health behaviours are consolidated and health paths can be altered positively or negatively; it is therefore a time when interventions may be particularly effective (Holmbeck, 2002). Negotiating the transition from adolescence into adulthood is highly significant for disease outcomes for the rest of an individual's life (Williams et al, 2002). As such it is critical that research examines young people's accounts of health and illness. Key factors identified as impacting on young people's experience of chronic illness include risk taking behaviour, autonomy development and positive adaptation or resilience.

Risk taking behaviours may include experimental behaviours that are often associated with adolescence such as risky sexual behaviour, drinking alcohol, taking drugs and smoking. Non-adherence to treatment and noncompliance with medical advice can also be seen as risk taking behaviour for young people with a chronic health condition. There is some evidence to suggest that young people with chronic health conditions are as likely (or more likely) than their peers to take part in risky health behaviours (Sawyer et al, 2007). Suris and Parera (2005) suggest that young people generally appear to perceive that everyone else is engaged in risk taking behaviour. They propose that chronically ill young people may be more susceptible than their peers to wanting to be like those around them in their eagerness to be 'normal' and thus more likely to engage in risk taking behaviours. However there have also been calls for a change in the way risk taking behaviour in adolescence is conceptualised, Michaud (2006) proposes; "*one of our crucial tasks is to advocate a positive attitude toward youth on the part of our colleagues and administrators, our politicians, the media, and the general public. Shifting the paradigm from risk-taking adolescents to adolescents who are exploring the world will enable us to advocate for youth from a positive position.*" (p483).

Changing the way in which we conceptualise risk taking behaviours by viewing them as signs of growing independence may empower young people, supporting the development of independence and autonomy. Autonomy is a psychological construct that emerges during adolescence; essentially this is a persons' ability to think about, feel and make decisions independently and is a particularly salient issue for many young people with a chronic condition (Holmbeck, 2002). During this life stage a shift occurs both psychologically and in practical terms as decision making moves from a parent to their child. Having a sense of autonomy may be an important factor in young people's wellbeing and positive management of a chronic health condition (Spear & Kulbok, 2004). The development of autonomy can impact on relationships between parents, young people and clinicians and can raise a number of ethical dilemmas that need due consideration in the provision of care for young people. Ethical issues are centred on confidentiality, disclosure, informed consent and power imbalance (Matutina, 2009; Kirk, 2007; Tan et al, 2007). As a young person matures they need age and developmental appropriate information and the opportunity to ask questions (Suris & Parera, 2005). Assuring confidentiality is crucial in providing healthcare to young people, yet concern about parental reactions places clinicians in a difficult position; during adolescence clinicians have to manage relationships with both parents and young people, heeding the wishes and needs of both (Berlen & Bravender, 2009; Jones & Bradley 2007). Another ethical consideration relates to informed consent and decision making in terms of treatment. British law is not clear on this issue; there is no completely clear cut-off point for consent with regards to age.

It is important to recognise that while chronic conditions can affect development, development can also affect chronic conditions. The relationship between developmental changes and chronic illness are complex and bi-directional (Suris et al, 2004). Psychosocial factors that appear to support young people's emotional wellbeing may include optimism, above average intelligence, social competencies, and positive relationship with a parent, family closeness and adequate rule setting by parents (Neinstein, 2001). Social aspects of life and the support of peers and friends have been highlighted as

supportive of positive adaptation in research with young people with a range of conditions (Reiter-Purtill & Noll, 2003; La Greca, 1990). The development of self-esteem and the formation of supportive coping mechanisms for life are formed during adolescence (Holmbeck, 2002), and this can be seen as a time of opportunity in which the development of positive coping and resilience can be supported (Ahern et al, 2008). Resilience can be thought of as either an outcome or as a process (Olsson et al, 2003). It tends to refer to the protective qualities and factors that support psychological adaptation and change. Factors are individual level resources (such as social skills, intelligence, self-efficacy, self-esteem) and societal processes including family dynamics and friends. These different protective factors are thought to support the development of resilience in young people. However the relationship between resilience, coping and chronic health conditions is unclear (Ahern et al, 2008). Research into how young people live with chronic conditions is crucial in order to understand the characteristics and processes that support the development of positive strategies that enable young people to live positively with a chronic health condition. In order to gain a holistic account of young people's experiences of managing a chronic health condition, research needs to recognise and explore positive adaptation and resilience as well as challenges and risk.

Knowledge and understanding of a chronic condition is important within management. This may be an important issue for many young people with NF1 (Rubinstein & Korf, 1990), particularly as a diagnosis may have been made when the young person was a young child, meaning that initial explanations about their condition may have taken place between health professionals and parents. Adolescence may also be a time when young people become increasingly aware of the potential to pass their condition on to future children. It is therefore important that they have up to date and correct information. Research with young people with other genetic conditions (including Epidermolysis Bullosa Simplex, Phenylketonuria and Congenital Adrenal Hyperplasia) suggests they want information about their condition and support in understanding it for themselves and in explaining it to others. In particular, visible aspects of genetic conditions can be difficult to explain to others (Williams et al, 2011; Szybowska et al, 2007). As such young people may therefore need support developing their understanding of their condition as they progress through childhood and into adolescence and adulthood.

It may be that the informational and care needs of young people with a genetic condition are similar to those with any other chronic health condition. However, there is little research evidence in this area and needs may be very different. Exploring young people's experiences of NF1 necessitates a consideration of the genetic nature of the condition which may have specific implications for young people. Genetic conditions can be particularly stigmatising (Peters et al, 2005; Chapple et al, 1995) and having a genetic condition during adolescence may make this time additionally stressful (Szybowska et al, 2007). Stigma is defined by Goffman (1963) as the way in which an individual is discredited and rejected because of a particular attribute. Since stigmatisation may disrupt positive adaptation and undermine resilience this may be an important aspect in young people's experience of NF1.

#### 1.4 The role of families

Families play a critical role in autonomy development and positive adaptation or resilience and can improve the emotional wellbeing of young people (Michaud et al, 2004) by supporting them in managing the transition from childhood to adulthood (WHO, 2013). Relations within families may be predictive of psychological outcomes for young people (Williams et al, 2002); family connectedness has been identified as important for young people's wellbeing (Wolman et al, 1994). Parents of a young person with a chronic illness may face many challenges. Times of transition or developmental milestones during adolescence may bring into focus an illness that is well managed day-to-day and, as such, may be particularly stressful. Parental coping has been associated with social support, the maintenance of normality, interpersonal resources such as stress management, as well as good information and understanding of the medical situation (Cavallo et al, 2009; Vermaes et al, 2008; Coffey, 2006; Hummelinck & Pollock 2005; Fisher, 2001).

Parenting young people with a chronic condition can be at odds with the developmental changes associated with adolescence. At this time young people generally move from a situation where parents are responsible for their health to taking responsibility for their own health and wellbeing (Holmbeck, 2002). Parents play a key role in supporting their child to develop the skills needed to autonomously and confidently manage their condition. Parenting that promotes the development of a young persons' autonomy, whilst remaining involved, appears to be highly supportive of positive adjustment and development (Willemen et al, 2011; Wong, 2008) whilst over protectiveness has been associated with lowered autonomy and risk of maladjustment (Holmbeck, 2002). Yet it is understandable that parents of a young person with a chronic illness may feel over protective towards their child and may be anxious about their growing independence.

While it is important to recognise that families play a crucial role in supporting young people to manage chronic illness in general, a genetic condition can impact on families in specific ways. As Williams (1994) writes "...*genetics is a speciality of medicine where awareness and consideration of the family is vital*" (p.55). On a medical level, many members of a family may already be diagnosed with the condition, or a diagnosis of NF1 in a child or young person may lead to further diagnosis within families. Care needs to be taken to examine ethical, psychological and practical issues.

Information about chronic illness is often addressed within family units. The manner in which young people are informed about and understand their condition is often reliant on their parents. In particular the communication of genetic information within families is often viewed as a family's own responsibility although support from health professionals may be sought and welcomed (Metcalf et al, 2011; Gaff et al, 2007; Forrest et al, 2003). Parents share information with their children with the aim of supporting their child's adaptation to the condition, based on their developmental needs, often waiting for children to ask questions (Gallo et al, 2005). Parents may need good quality and up to date information to help them communicate with their children about genetic conditions (Metcalf et al, 2008; 2011; Gallo et al, 2005). However the way in which individuals and families understand and interpret genetic conditions may not just be related to medical knowledge and information. Families

build their own narratives; lay understandings of genetic inheritance can be dominant, meaning that medical knowledge can be viewed as one kind of knowledge whilst families evaluate their own understandings of the condition. Santos and Bizzo (2003) found that some families with NF1 thought the condition was associated with a relative with syphilis, while others reported a link between skin colour and NF1. In addition, some parents may appreciate uncertainty within genetic information; it may be a useful coping mechanism (Whitmarsh et al, 2007).

### **1.5 The role of health professionals and healthcare systems**

During adolescence and early adulthood, as well as changing family relationships, long standing relationships with health professionals may end and new relationships begin as young people transition from paediatrics into adult health care. Health professionals play a key role in supporting both parents and young people throughout childhood into adulthood in many diverse ways. For instance health professionals' interactions with education systems have been highlighted as supportive of young people with chronic illness (Suris, 2004) and as children and young people with NF1 may have associated learning difficulties, this may be highly significant support for these young people and their parents. Additionally, parents report wanting help and support from clinicians in discussing and informing their children about genetic conditions (Gallo et al, 2005). Informational needs, particularly around a genetic condition, change through childhood into adolescence and adulthood (Metcalf et al, 2011). Supporting parents in providing this information and keeping both parents and young people informed of developments is an important factor in delivering healthcare to young people.

Transition from adolescence into early adulthood is a significant time for the development of self reliance for young people with a chronic condition. In practical terms, transition between paediatric and adult care needs to be well managed (Jones & Bradley, 2007) and recognised as a significant life event for young people (Por et al, 2004). Whilst the importance of transition is generally recognised (DoH, 2008) the management of transition is inconsistent and young people report that the process is challenging, often abrupt and a time of distress and anxiety (DoH, 2008; Maunder, 2004). The impact of poor management of transition can be severe; Kipps et al (2002) report young people's attendance within diabetes services dropped from 94% before transition to 57% two years after transition. For young people it can be difficult to move from a service that feels familiar and comfortable into adult care which feels unfamiliar and less friendly (McDonagh & Viner, 2006; Fleming et al, 2002). Negative experiences can increase the risk of disengagement with healthcare services at a time when the development of positive engagement with healthcare is imperative.

Transition to adult care means that young people may be seen in new clinics, or new hospitals, or may see the same team but as an adult patient. In the case of NF1, in the UK, young people without complex needs may change from seeing a specialist or paediatrician to being referred to primary care and will be expected to arrange an appointment with their General Practitioner once a year for a check up (Ferner et al, 2007). However, regardless of physical and structural changes, relationships

with health professionals alter both in terms of expectations placed on the young person and the involvement of parents. Pediatric care is family centered while adult services emphasize independence and self-management, meaning adult clinics can feel intimidating for young people (McDonagh & Viner, 2006; Viner, 1999,). Transition requires a change in self-perception which some young people may find difficult (Viner, 1999).

Health professionals play a key role in supporting and managing the transition of young people from paediatric to adult care yet for many health professionals transition and working with young people may not have been part of their training and they may feel they lack expertise or confidence in meeting their particular needs (DoH, 2007). Working with young people can be seen as a particular skill and one that both paediatric and adult staff may be wary of. Even among staff who are aware of young people's issues and enjoy working with young people meeting the needs of large numbers of adults takes precedence if transition programmes are not well established (Fleming et al, 2002). If systems are not in place to manage transition, then it can be a time when no single team or person, neither paediatric nor adult services, feels responsible for the young person's transition (RCN advice for nursing staff, 2004).

However, most transitions are successful and many young people look forward to adult care (Madge & Bryon, 2002; Nasr, 1992). Indeed transition can be an opportunity for health professionals to ensure that young people are well informed about healthcare in general and their condition specifically. As Gorter summarizes; "*Working with adolescents and their families in (clinical) transition is exciting, challenging, and sometimes difficult and frustrating. It also is an ideal time to educate adolescents about how to best care for their bodies and their life.*" (Gorter, 2009 p.365).

## **1.6 Conclusion**

This chapter has described NF1 and defined how this thesis interprets the term young people. Following this it has highlighted that there are a range of influences that may impact on how young people adapt to and live with a chronic condition such as NF1. Adolescence and early adulthood is undoubtedly a crucial developmental stage with regards to health and wellbeing. This is a time during which interventions and support may be highly beneficial. Supporting young people to adapt positively to a chronic condition during the transition from adolescence into adulthood may be highly supportive of lifelong positive management of a chronic condition. Chapter 2 now moves on to specifically consider the psychosocial impact of NF1 during adolescence and the importance of attending to the role of appearance within this.

## **Chapter 2: Literature review**

The current chapter focuses on reviewing the literature regarding the psychosocial impact of NF1 and then reviews the literature related to the importance of appearance and the factors involved in living with an altered appearance. Initially literature was reviewed widely in order to gain a broad overview of these key areas; this involved searching relevant databases (including PubMed, PsycINFO, JSTOR, EBSCO, MEDLINE and CINAHL) for literature related to NF1, appearance and visible difference. As there is limited literature regarding the psychosocial impact of NF1 literature in this area was reviewed in a systematic manner and was regularly updated throughout the research process by searching for key words 'NF1' and 'neurofibromatosis' since the date of the previous search. Literature relating to visible difference was reviewed by concentrating on the psychological management of and adjustment to a visible difference and specifically research with young people with a visible difference, again this review was updated regularly. In addition as the research was conducted findings pointed to new areas of literature, these are reviewed in subsequent chapters.

### **2.1 The psychosocial impact of NF1**

NF1 cannot be cured; one of the main focuses for clinicians therefore is supporting adaptation to the condition (Rubenstein & Korf, 1990). Adaptation refers to the process of adjusting to change (either actual physical change or new information). In the current context, psychological adjustment reflects how individuals manage the impact of NF1 on their wellbeing. In order to support positive adjustment it is essential to understand the potential psychosocial impact of NF1 on individuals throughout their lives and the day-to-day experiences of those affected. However there is little research that specifically examines the psychosocial aspects of the condition. The following sections consider current knowledge regarding the psychological impact of NF1 on children, young people and adults.

#### **2.1.1 Psychosocial experience of children and young people with NF1**

The limited literature that has examined NF1 in childhood and adolescence has explored the impact of learning and behavioural difficulties including Attention Deficit Hyperactivity Disorder (ADHD) and Autistic Spectrum Disorders (ASD), quality of life, the role of the self-concept, as well as social, emotional and behavioural difficulties.

Up to 65% of children with NF1 may have learning difficulties (Cutting et al, 2004), most frequently relating to literacy and cognitive difficulties. It has been found to have a significant impact on school performance (Ferner et al, 2012; Krab et al, 2008). Dilts et al (1996) propose there is a group of children with NF1 who are at risk of academic problems; they may be under identified in schools, possibly due to a lack of awareness of NF1 and the learning difficulties often associated with the condition. Academic achievement of children with NF1 may be broadly affected through different learning difficulties, behavioural concerns, and attentional deficits. Behavioural problems, including inattentive behaviours, are commonly reported and children with NF1 can appear more anxious and depressed than their unaffected peers (Payne et al, 2011; Hyman et al, 2006; Descheemaeker et al, 2005; Johnson et al, 1999; Brewer et al, 1997; Dilts et al, 1996; Counterman et al, 1995). Parents

describe their children with NF1 as being shy, internalising problems, finding interactions with peers difficult (Johnson et al, 1999; North et al, 1997; Dilts et al, 1996) and appearing to have poor self-awareness (Descheemaeker et al, 2005).

Symptoms of ADHD and ASD are more common in children with NF1 than in the general population (Ferner et al, 2007; Williams & Hersh, 1998; North et al, 1997; Spaepen et al, 1992). Up to 50% of people with NF1 meet the criteria for diagnosis of ADHD (Gilboa et al, 2011) and its presence may undermine the academic achievement of children with NF1 (Pride et al, 2012) as well as social functioning (Barton & North, 2004). However attentional problems may not be ADHD specific, children with NF1 have been found to have deficits in attention even without the presence of ADHD (Isenberg et al, 2013). A recent study (Garg et al, 2012) found a high prevalence of ASD symptoms in children with NF1 reporting 30% prevalence within clinical range (based on parental reports) while a further 30% scored in the mild-moderate range of ASD symptoms. The study also found the majority of children who met the ASD criteria also met the ADHD criteria. However, while it is well reported that children with NF1 have a range of cognitive and behavioural deficits, there is little explanation of how this impacts on day-to-day life.

Much of the research examining the overall psychosocial impact of NF1 on children and adolescents has focused on quality of life (QoL), this is a multifaceted concept that measures the entirety of factors that may impact on a person's life. The World Health Organisation (Sheiham, 2005; Kuyken et al, 1995) defines these domains as (1) Physical, (2) Psychological (including positive feelings, thinking, memory, learning, concentration, self-esteem, body image and appearance and negative feelings), (3) Level of independence, (4) Social, (5) Environmental and (6) Spiritual (religious, and personal beliefs). These domains are generally assessed using questionnaires or interviews. Quality of life gained attention after parental reports of concerns related to the impact of NF1 on social skills and relationships (Kayl & Moore, 2000) and NF1 has been found to have a significant impact on QoL in many domains and using different measures (Krab et al, 2009; Wolkenstein et al, 2009; 2001; Noll, 2007; Oostenbrink et al, 2007; Graf et al, 2006; Page et al, 2006).

QoL has been examined in children with NF1 of varying ages. In a study of very young children (12-72 months) Oostenbrink used parental reports of health related QoL (HRQOL includes self-reported measures of physical and mental health) and demonstrated that perceived severity, complications, visibility of NF1, familial NF1 and educational level of parent all impacted on the ITQOL (Infant and Toddler QoL). Similar findings by Wolkenstein et al with older children aged 8-16 (and parental reports) also demonstrate that NF1 has a strong negative impact on QoL, particularly in terms of social inclusion, exclusion and independence. Parents rated their child's QoL lower than the children themselves did, although this was less in familial cases of NF1. It could be argued that parents' pessimism on these measures, indicated by lower scores on QoL measures, could in itself lead to potential issues around social inclusion, exclusion and independence for the young people. The effect of families on QoL was also investigated by Graf et al (2006) through standardised interviews with children and young people aged 7-16 (mean age of 11.6 years) and parental questionnaires. QoL of children and young people was found to be impaired and psychological adjustment (measured by

the Child Behaviour Checklist) disturbed. The authors report greater internalising behaviours (such as social withdrawal, and anxiety) and externalising behaviours (including aggressive behaviours). High family cohesion and low conflict were found to be positively associated with parental reports of QoL and psychological adjustment in children and young people with NF1. A further study by Krab et al (2009) reported that NF1 had a profound impact on social, emotional, physical and behavioural aspects of health related QoL. However, as with Wolkenstein's findings, parents were generally more pessimistic than their children, particularly in their rating of NF1 on family life and on them as parents, as assessed by way of the Child Health Questionnaire. Findings that parents', children's and young people's accounts differ are common within the wider literature relating to quality of life measurement in children (Eiser & Morse, 2001). It may be that parental reports highlight their anxiety related to their child's condition rather than the child's actual quality of life.

In an investigation of social, emotional and behavioural quality of life, Noll et al (2007) compared 58 children with NF1, aged 7-15, with non-affected peers. Children were evaluated by researchers, parents, teachers, peers and self report. In line with other studies (eg Barton & North, 2004; Johnson et al, 1999; Dilts et al, 1996) parents and teachers reported children with NF1 as having some social difficulties. However this study went further in clarifying the domains in which social problems occur. Peers and teachers suggested children with NF1 are more sensitive and isolated; they had fewer friendships and were less well liked by their peers. Parents of the two groups did not report great differences in their child's emotional well being although parents of children with NF1 reported greater social difficulties than parents of unaffected peers. In terms of behaviour, peers did not report aggression and disruption. Disease severity and psychological functioning were not strongly associated, and physical appearance was not significantly related to any of the psychological measures used. However appearance was measured objectively by a research assistant and through examining medical charts. As such, objective severity was being measured rather than individuals' subjective accounts of their appearance, which may be more relevant to psychological experience of NF1 (Ferner, 2012).

The quality of life studies outlined in this section highlight the social and emotional impact that NF1 can have on children and young people. It is important to note however that QoL is difficult to define and to measure. A NF1 specific QoL measure for adults has recently been developed (Nutakki et al, 2013) and this may capture the HRQoL of individuals with NF1 more precisely (domains/sub scales include physical, emotional, social and cognitive functioning, communication, worry, perceived physical appearance, pain and hurt, paresthesias, skin irritation, sensation, movement and balance, daily activities, fatigue, treatment anxiety and sexual functioning). Versions for young people and parents are also reported as being in development (Nutakki et al, 2013).

In addition to investigating quality of life, the social and emotional concerns of children and young people with NF1 have been specifically examined in terms of the self concept (Barton & North, 2007), social skills (Barton & North, 2004), social information processing and cognition (Huijbregts & Sonnevile, 2011; Huijbregts et al, 2010; Johnson et al, 1999). The impact of NF1 on the self concept (how an individual perceives and evaluates themselves) and social skills generally is unclear. Dilts et

al (1996) describe how parental reports suggest that their children with NF1 did not differ significantly from non-affected siblings on measures relating to social skills, personal living skills or self concept. However parental reports and children's own reports have been found to differ, with children reporting lowered self worth and concerns about their behaviour while parents were more positive about their child's adjustment (Counterman et al, 1995). Using self report measures Barton and North (2007) found children and young people reported a positive self concept, although they are reported as likely to develop a negative self concept specifically in relation to physical and sporting abilities. However again parental reports and the reports of young people differ and sibling's scores were similar to the children and young people with NF1 which could suggest that any impact on the self-concept is more related to familial variables than NF1 specifically.

While children and young people with NF1 have been identified as having difficulties with social relationships (Sebold et al, 2004; Johnson et al, 1999; Dilts et al, 1996; Benjamin et al, 1993), few studies have specifically examined the social skills and social outcomes of children with NF1 or identified why they may be impaired. In their examination of social skills of children with NF1, Barton and North (2004) highlight ADHD as a major risk factor, suggesting it is not NF1 alone that affects social skills but rather having both NF1 and ADHD. However, in their study investigating social information processing in children and adolescents with NF1, Huijbregts et al (2010) found that even when excluding participants with ADHD there were still differences suggesting there may be ADHD and NF1 specific deficits. In particular, Huijbregts et al (2010) found individuals with NF1 have problems processing social information both cognitively and in terms of recognising social signals; in a face recognition task children and adolescents with NF1 had difficulties recognising fear and anger.

Both ADHD and ASD can impact upon language skills and development, and children with NF1 and ADHD have been found to perform significantly more poorly on receptive language (the comprehension of language which can impact on understanding complicated sentences and spoken instructions) (Pride et al, 2012). People with NF1 may be at risk of poor language skills; difficulties with articulation and fluency have been reported in children and adults (Cosyns et al, 2012; 2010a; 2010b) while speech and writing tasks have been found to be impaired in adults (Lorch et al, 1999). The high prevalence of ASD, ADHD and specific language difficulties, alongside reported social emotional and behavioural difficulties may impact on the social skills and interactions of children and young people with NF1. In order to understand the social competence of young people with NF1 a greater understanding of social information processing may be beneficial. Lehtonen et al (2013) suggest that, within this, attention should be paid to understanding gestures and tone of voice as well as higher order processes.

A 'behavioural phenotype' for children with NF1 was suggested by Dilts et al (1996) after comparing 20 6-17 year olds (mean age of 10 years 10 months) with age and sex matched siblings. Social problems associated with anxiety, depression and psychosocial concerns were found to be linked to social acceptance, particularly relationships with peers. Children and young people with NF1 were found to often lack peer support. In particular the authors proposed that competencies, appearance and physical disease factors interact with the social context, and that these interactions contribute to

the impact of NF1. The behavioural or psychosocial aspects of NF1 are highlighted as crucial in determining the overall impact of the condition. Kayl and Moore (2000) support the view that children and adolescents with NF1 have a behavioural phenotype, suggesting that this includes specific cognitive and learning deficits, behaviours characteristic of ADHD and limitations in social functioning including social awkwardness. They write that children with NF1 often seem to be teased; not simply due to the physical signs of NF1 but also because of poor social and interpersonal skills. However, they also acknowledge that physical and behavioural manifestations are highly variable and that it is unclear whether problems relate to influence at a genetic level or internal and external perceptions of NF1. Prinzie et al (2003) found the personality profile of children and adolescents with NF1 appears to be different to those without it. In particular, those with NF1 appear less conscientious, less emotionally stable, less open to new experience, have less motor activity, and appear more extrovert, dependent, and irritable yet equally agreeable. Findings were similar in familial and de novo NF1 and no association was found with gender, severity of NF1 (both medical and cosmetic severity, as rated by researchers) or IQ. However within this, as with Kayl and Moore (2000), the authors also report individual differences, suggesting it is important to bear in mind the variability of NF1. A recent systematic review of the behaviour of children with NF1 (Lehtonen et al, 2013) concluded that *“Overall, children with NF1 appear to display higher frequency of different emotional and behavioural problem symptomatology than comparison children or normative samples”* (p.121) and suggests that more work is needed to understand the psychosocial challenges associated with NF1.

The literature outlined in this section demonstrates that NF1 can impact on children and young people socially, emotionally and behaviourally as well as on their overall quality of life. Psychological adjustment may be impaired and family variables are significant, yet reports by parents and children vary (Krab et al, 2009; Oostenbrink et al, 2007; Graf et al, 2006). Children may be socially isolated (Noll et al, 2007) and NF1 related variables impact emotionally (Graf et al, 2006). It would appear that defining characteristics of children and young people with NF1 may be an unhelpful approach; NF1 is characteristically variable and unpredictable. Describing the variety of the experiences of young people and finding ways to support and facilitate positive adjustment may be more useful than attempting to define the characteristics of those with the condition.

### **2.1.2 Psychosocial experience of adults with NF1**

Although this thesis concentrates on young people’s experiences during adolescence and into early adulthood, it is useful to review how adults experience NF1 in order to consider which aspects may become important as young people move through adolescence. While the research reviewed in the previous section has a particular focus on social and behavioural characteristics of children with NF1, research with adults with NF1 has found managing an altered appearance to be a key theme. The emergence of appearance as a theme within research with adults may reflect physical appearance changes associated with NF1 during adolescence, or it may reflect a change in how individuals perceive their appearance. However, it may in part reflect different questions asked in research with children and young people and with adults.

Samuelsson and Samuelsson (1989) report complex psychosocial concerns associated with NF1, including concerns related to appearance. Participants reported avoiding undressing in communal changing rooms, not sunbathing, covering up and not wanting to wear bathing suits because they felt uncomfortable with others seeing and commenting on their neurofibromas. Nine participants were particularly distressed by questions from others about their neurofibromas and avoided showing them. However, no significant positive relationship between the physical severity of NF1 and mental illness was found. A 12 year follow up of adults in this study (Zoller & Remback, 1999) found the impact of NF1 on participants continued but did not progress. Reviewing the psychological aspects of NF1 amongst adults, Mouridsen and Sorensen (1995) also identified appearance concerns as significant, they suggest this is partly due to the high value society places on physical attractiveness and also as people with NF1 may face prejudice. Furthermore the authors propose that the unpredictability of NF1 means that “....*even a patient with minimal manifestations of the illness may live in constant fear and uncertainty*” (p.921), indicating that severity of NF1 may not determine psychological adjustment.

Ablon's (1999) anthropological study of the psychosocial impact of NF1 explored how the condition impacts on daily life. Her work involved interviews with adults and families with children with the condition over an eight year period, and examined the lived experience of individuals with NF1. In her book she writes “*the severe impact of NF1 on the lives of the many subjects was apparent throughout the interviews.....social stigma and rejection appear to exact a far more frequent psychological and social price on subjects than do physical symptoms*” (p143). Ablon found a common concern for people with NF1 was the uncertainty of the condition, which related to both medical concerns and appearance concerns. Although Ablon did not directly interview children and young people she reports that adults retrospectively describe adolescence as a challenging time, social concerns and appearance concerns were common and participants described feeling different and isolated. Ablon also emphasises that the diversity of symptoms may mean there is a lack of commonality between people with the condition. Ablon's work highlights that there may not be a unified NF1 identity, a finding supported by more recent research with adults with NF1 (Carrieri, 2011) which has suggested that this lack of a unified identity may impact on how people perceive their condition.

In their study investigating coping in adults with NF1 (Dheensa & Williams, 2009) described participants feeling judged by others in relation to the way they looked and reported a lack of social support. Despite recommendations that people should have clear and accurate information about their condition in order to adapt positively (eg Ferner et al, 2007), participants felt there was insufficient information available for them. The authors highlight the paucity of research into the psychosocial needs of people with NF1 and suggest there is a need to focus on supporting people with NF1 to develop positive coping strategies. Such strategies may support and develop individuals' understanding of their condition as well as their confidence in discussing NF1 with others in order to gain social support and the understanding of family and peers.

Whether or not to tell others about their diagnosis was identified by Hummelvoll and Antonsen (2013) as a particular concern for many participants in their interviews with adults aged 18-37 with NF1. In addition to having clear information about their condition, they suggest that people may need support

in explaining NF1 and talking to others about it. The study also highlighted that low self confidence affected many of the participants and unpredictability was again identified as a key concern. In line with findings from earlier studies (including Mouridsen & Sorensen, 1995; Samuelsson & Samuelsson, 1989) the authors suggest that psychosocial factors appeared to impact on individuals more than severity of NF1. This finding is further supported by Ferner (2012) who writes that an individual's self perception of their appearance in relation to NF1 may not be the same as an observer's objective perception of them; while some people with barely perceptible neurofibromas suffer anxiety and depression, others with a high tumor load cope well.

The impact of the visibility of NF1 symptoms on body image has been investigated specifically by Granstorm et al (2012) whose survey found that adult NF1 patients had a negative body image. Within this, the impact of disease visibility was mediated by how individuals experienced their body, suggesting that general views about the body and appearance are an important aspect to consider, and may be more relevant to psychosocial adjustment than the visible aspects of NF1. Smith et al (2013) reported that the majority of women with NF1 in their study indicated appearance related concerns. Two thirds of these concerns were described as NF1 related including concerns regarding cafe-au-lait spots and tumors. A third of appearance concerns were more general regarding other issues including height and weight. Women with NF1 reported significantly higher levels of social self consciousness than both population norms and women who had undergone a mastectomy. The authors highlight that appearance concerns, social self consciousness and lowered self esteem may all impact on the quality of life of women with NF1.

Appearance is clearly important to adults with NF1 and the literature reviewed in this section highlights that it is its impact on social interactions including explaining NF1 to others and concerns about how others might react, rather than the severity or visibility of the condition that may be particularly salient. The following section considers the importance of appearance and how people adjust to a visible difference before concentrating on the impact of visible difference during adolescence. The chapter then specifically considers what the current literature tells us about young people, appearance and NF1.

## **2.2 Appearance**

### **2.2.1 The importance of appearance**

Physical appearance is generally highly valued within society. This is not a new phenomenon, it has been valued across cultures and history, although appearance concern is described as now having reached 'epidemic proportions' (Rumsey & Harcourt, 2005). Appearance concerns affect people of all ages; 40–50% of 6–12 year olds report being unhappy with their appearance (Smolak, 2011), to the extent that dissatisfaction with one's body can be classed as 'normative discontent' (Rodin et al, 1985). "What is beautiful is good" is a strong and prevailing stereotype (Dion et al, 1972). The media presents being young, thin and attractive as good; being unattractive or large is a negative trait that

can be improved, through dieting, exercise or investing in one's appearance (Halliwell & Diedrich, 2012). The development of body image (how individuals perceive and feel about their body) comprises biological, socio cultural and psychological factors (Cash & Pruzinsky, 2004). Socio cultural influences on body image include the messages received from family, peers and the media and the internalisation of these ideals can become part of an individual's body image (McCabe & Ricciardelli, 2001). It is important to note that body image concerns relate both to individuals' dissatisfaction with their own body, and also to their view of how others regard their body (Davidson & McCabe, 2006). Body image is probably the most important component of global self esteem during adolescence (Cash, 2004) and is relevant to boys and girls (Davidson & McCabe, 2006). This focus during adolescence is due in part to the physical changes that occur during puberty which mean that body shapes alter. Compounding this, adolescence is a time during which socio cultural messages about appearance are particularly strong. Media images play an important part in the conceptualisation of body image; children and young people may be especially vulnerable to this (Dohnt & Tiggemann, 2006). Messages that young people receive from school, peers, family and the media have been found to play a critical role in shaping their body image (Gillen & Lefkowitz, 2009). During adolescence there is a heightened self awareness and an increased awareness of others' perceptions of their bodies (Blakemore, 2008). Late adolescence to early adulthood appears to be a period of increased appearance salience (meaning that appearance becomes more important to individuals) and concern (Moss & Rosser, 2008). Seventy percent of adolescent girls and 45% of boys report that they want to change their body weight or shape (Smolak, 2012).

### **2.2.2 Adjustment to visible difference**

Against this backdrop, where appearance concern is so prevalent, what might it mean to have a visibly different appearance? Over one million people in the UK have a significant disfigurement to the face or body (Changing Faces, 2012). It is difficult to define what is or isn't a visible difference as individuals' perceptions and evaluations of their appearance are highly subjective; what may be considered irrelevant to one person can be highly salient for another. Appearance can be altered by health conditions, treatment or trauma. Visible differences can be acquired (through accidents, injuries, or illness), congenital (including cleft lip and palate, birthmarks and scoliosis) or genetic (including NF1).

Psychological adjustment to a visible difference is a highly individual process, whilst some people adjust and live positively, others find adjustment challenging. A visible difference can have a profound psychological impact on an individual, in terms of their body image, quality of life, self esteem and self concept (Rumsey & Harcourt, 2005; Moss & Carr, 2004; Kent & Keohane, 2001; Thompson & Kent, 2001; Newell, 2000). There are a range of individual factors involved in adjustment to a visible difference. Newell (1999; 1998; 1991) suggests that while avoidance and fear of social situations following an altered appearance are common, an individual's response is mediated by life events, personality, history of changes to body image, body image coping strategies, fear of the changed body and the reactions of others.

The reaction between the social context and individual processes has been highlighted as an important factor in adjustment (Thompson & Kent, 2001). Partridge and Pearson (2008) suggest that psychosocial distress for people with a visible difference has two main causes, first interpersonal difficulties which relate to social situations and other people's reactions and second, intrapersonal difficulties which relate to living in a society in which the "beauty is good" stereotype prevails. Day to day challenges facing people with a visible difference often relate to managing the reactions of others, including dealing with staring and being asked questions (Lansdown et al, 1997). Social avoidance, distress, fear of negative evaluation (concern about being judged negatively by others) and feelings of shame have been identified as key factors in adjustment to a visible difference (Moss & Rosser, 2008).

The visible signs of a condition may therefore lead to stigma experiences. Stigma impacts on self-esteem, academic performance and health (Major & O'Brien, 2005) and it is important to note that this relates not only to actual experience and the actions of others but also to individuals' expectations of encountering stigma. Scambler and Hopkins (1986) suggest that stigma can be 'enacted' or 'felt'. Felt stigma relates to the fear of being stigmatized whereas enacted stigma is the actual experience of facing discrimination. Furthermore Scambler and Hopkins suggest felt stigma may present more psychological challenge than actual enacted stigma. In an appearance context this suggests that individual's concerns about possible stigma are highly relevant regardless of actual stigma experience. Enacted stigma is widely reported by people with a visible difference (Lovegrove & Rumsey, 2005; Thompson & Kent, 2001; Thompson et al, 2002). Supporting those who feel stigmatised may involve understanding resilience as well as vulnerability (Major & O'Brien, 2005).

It is crucial to recognise that an individual's own subjective perception of their difference is the key to understanding their psychological adaptation (Hansen & Clarke, 2008). This is described as an ongoing process, part of which is informed by the value placed on appearance by an individual which appears to be a highly significant factor within adjustment. Self perceptions of appearance are critical in adjusting to an altered appearance (Moss & Rosser, 2012). These perceptions are stored and organised within the self concept, and are subject to cognitive biases in the processing and storage of appearance related information and encounters. For instance people with high levels of appearance concern may attend to, interpret and remember appearance related information and events more readily which in turn reinforces concerns. These processing biases may not cause appearance related anxiety but may increase an individual's focus on appearance. Furthermore, as these processes are related to subjective evaluations, this could suggest that having an elevated awareness of appearance could be a risk factor for poor adjustment regardless of the actual noticeability or objectively judged severity of any visible difference. Research has demonstrated that the self concept of participants identified as poor adjusters was found to be organised so that appearance information was more important to them (Moss & Carr, 2004). Therefore the salience of appearance may be an important factor in adjustment to an altered appearance; if an individual places a high value on

appearance they are likely to experience greater distress relating to visible difference (Lawrence, 2006a).

While the salience of appearance may be an important factor within adjustment, it is important to reiterate that levels of appearance concern are not necessarily related to the nature or severity of a visible difference. There is no obvious relationship between severity of a disfigurement and extent of psychosocial adjustment (Moss, 2005). A very noticeable visible difference may result in predictable reactions from others, for instance a birthmark on a person's face that is highly noticeable may lead to regular comments and questions about the mark to which the person can provide a practiced answer. In contrast, an area that is less visible or noticeable to others may result in increased efforts to conceal the difference leading, therefore, to increased anxiety related to revealing a visible difference. For instance, a birthmark on the back may only be noticeable when wearing a swimming costume or when getting changed and as such is rarely commented on or noticed meaning that explanations are rarely given and therefore may be more anxiety provoking when they are required. It seems that small concealable differences may cause similar psychological discomfort as more obvious unconcealable differences (Moss & Carr, 2004). It is also important to note that objective and subjective measures of severity may not correlate (Moss, 2005). It is individual, subjective assessment of severity that is critical in adjusting to a visible difference (Ong, 2007) reflecting the individual and subjective nature of body satisfaction-dissatisfaction, a point also raised in observations of clinical experience of working with people with NF1 (including Ferner et al, 2012).

In considering adjustment to a visible difference it is important to resist a pathologising approach. If it is assumed that experience is negative, then positive experiences may be missed. Living positively with a visible difference has been shown to involve, in particular, a strong sense of identity, positive social support and strong social skills (Clarke, 2008; 1999; Rumsey & Harcourt, 2005; Moss & Carr, 2004; Rumsey et al, 2004; Kent & Keohane, 2001; Thompson & Kent, 2001). A large scale mixed method research programme (n=1265) examining the psychosocial aspects of an altered appearance in adults (Rumsey et al, 2010) concluded that positive adjustment to a visible difference is associated with higher levels of optimism and social support, low concerns about others evaluations, lowered importance placed on appearance and feeling able to disguise the difference. Individuals' management of a visible difference is a highly subjective experience and how they manage and process situations will depend on previous experience, individual body image and psychological processes which in turn have been influenced by previous social situations (Kent & Keohane, 2001).

Appearance changes associated with NF1 are variable and unpredictable meaning adjustment needs to be dynamic and may be a lifelong process. Rather than adjusting to a change in their appearance, people with NF1 need to be able to manage both uncertainty and actual changes on an on-going basis. The nature and unpredictability of NF1 suggests resilience may be a critical factor. Resilience can be thought of as a force field around a person (Bradbury, 2007); in terms of visible difference it has been defined as having the self confidence to withstand social and psychological pressures (Rumsey & Harcourt, 2005). Understanding resilience necessitates examining risk factors and

positive adjustment (Feragen, 2012). An understanding of these factors can then be used to develop appropriate support and intervention. In order to identify risk, positive adjustment and resilience it is critical to understand how a visible difference impacts on an individual day-to-day, both positively and negatively.

### **2.2.3 Young people and visible difference**

Appearance concerns affect the daily life of most young people (Rumsey & Harcourt, 2005). Having a condition that alters their appearance increases the risk of negative effects on psychological well being (Rumsey & Harcourt, 2004). Young people may have appearance concerns around standing out and fear of not fitting in with their peers. As conformity becomes increasingly important during adolescence, having an existing visible difference may become more challenging (Griffiths, 2012) whilst it may also be a particularly difficult time to acquire a visibly different appearance (Ben-Tovim & Walker, 1995). Young people with a visible difference have reported teasing, bullying and stigma (Strauss et al, 2007; Lovegrove & Rumsey, 2005), they may have an increased risk of developing appearance concerns and may be at risk of psychosocial maladjustment (Benrud-Larson et al, 2003). However, research also demonstrates that young people can be exceptionally resilient to appearance changes; many young people adapt well (Rumsey & Harcourt, 2007; Wallace et al, 2007).

As outlined in the previous section social skills are significant in adjustment to a visible difference. This is not only in terms of mediating social interactions, but also in terms of the way in which young people internalise and manage others' reactions to them (Feragen et al, 2010). Adolescence is a stage during which social relationships become more prominent leading to suggestions that social encounters in adolescence have long lasting importance for young people with a visible difference (Fox et al, 2007). Young people may fear negative evaluation from others which can lead to social avoidance exacerbating interpersonal difficulties and impeding the development of social skills. This then returns full circle and the resulting impact on social skills can in itself cause distance from peers and possible social isolation (Chamlin, 2006; Tan, 2004).

While there are certainly challenges within the social realm for young people with a visible difference, their social world can also be a key to resilience. It is important to consider the positive role of friendships and social interactions. Social support is fundamental to positive adjustment to a visible difference and many young people adjust positively to changes and display resilience (eg- Griffiths et al, 2012; Feragen et al, 2010 a; b; Williamson et al, 2010; Wallace, 2007). With the increased importance of social relationships during adolescence, positive interactions at this time may support long term positive social skills and strong social support, leading to positive self belief. Friendships, in addition to supporting the development of social skills and social support, can be a buffer against internalising and negative adjustment (Egan et al, 2011; Dovey-Pearce et al, 2010; Feragen et al, 2010 a; b; Thompson & Broom, 2009; Rumsey et al, 2004; Thompson & Kent, 2001). Social experience informs a young person's internal self perceptions, and an association has been

demonstrated between social interactions and subjective appearance evaluations (Feragen et al, 2010). Positive social encounters and friendships during adolescence may support emotional resilience.

In order to understand how social encounters impact on young people, research needs to focus on how young people 'live with' visible difference, concentrating on describing and theorising positive resilience (eg Feragen, 2010; Prior & O'Dell, 2009; Wallace, 2007). There is now an emerging and growing body of work examining young people's day-to-day experiences of living with a visibly different appearance. For instance, Larouche and Chin-Peuckert (2006) explored the body image of five young people during cancer treatment. The authors describe how participants discussed feeling exposed and talked of people looking at them. In terms of how body image disruption impacted on their lives, young people discussed avoiding social situations and attempting to maintain normality through minimising changes by using clothes and make up. The term 'peer shielding' is used by the authors to describe how friends were supportive, especially when going out; they provided comfort and protection. Some participants talked of testing the waters and checking how they looked with friends. This study draws attention to the feelings of difference and vulnerability that were expressed by the young people. However it also highlights the positive role of friends in shielding and supporting social activities and reintegration. The importance of friends in managing a visibly different appearance is further highlighted by Fox et al (2007) who employed grounded theory to analyse group discussions between young people with psoriasis. Eight young people aged between 11 and 18 years took part in online focus groups and talked about interactions with a variety of others including medical professionals, friends and peers. Fox et al discuss a strong sense of struggle for the young people in the study which impacted on self confidence, social functioning and perceived stigma, which were mediated by friends and social support.

Wallace et al (2007a) interviewed six females aged 14-19 who had completed treatment for cancer within the previous two years. These in-depth interviews were analysed using interpretive phenomenological analysis (IPA) which identified appearance concerns as highly important to all participants; in particular hair loss was identified as the worst part of their treatment. The young people managed changes to their appearance by using practical strategies such as cutting their hair or wearing hats. Supporting Larouche and Chin-Peuckert's (2006) findings, young people had a strong desire to appear 'normal' to others and the appearance changes that occurred were seen as a sign of the illness and change. Wallace found a shift in the meaning of appearance after cancer; many aspects of their appearance were reinterpreted as more positive after having had a changed appearance, they placed less value on appearance and had a greater appreciation of life in general. In a second study by Wallace et al (2007b) appearance changes were also found to be central to young people's experience of meningococcal septicaemia (MS). Wallace interviewed 11 young people in depth and again interviews were analysed using IPA. Young people reflected that their lives were different before and after MS. Again participants wanted to achieve 'normality'. Social comparison was used by participants, initially upwards by comparing themselves to others who did not have an altered appearance and later downwards toward others who appeared to have a more

altered appearance than themselves, supporting their own adjustment. Participants felt differently about scarring and appearance changes than their healthcare professionals did and successful treatment was viewed differently in that health professionals were happy with wounds healing while participants were distressed by scars. Wallace suggests that this may relate to both parties making different comparisons; health professionals may compare to more severe patients whilst young people are comparing themselves either to others or to their own body before changes occurred.

Overall, the two studies by Wallace et al (2007 a & b) demonstrate the impact that appearance changes during adolescence can have on self esteem, behaviour, social interactions, the importance of appearance to the young people, the subjective nature of concerns and comparisons made with others. Whilst changes to appearance had caused disruption and been distressing for individuals they had mostly adjusted positively, however interviews took place some time after changes to appearance had occurred, meaning young people had time to adjust. Additionally, it may be likely that individuals who have adjusted well are more likely to want to participate in research about their experience. As in the previous studies discussed in this section (Fox, 2007; Larouche & Chin-Peuckert, 2006) social interactions and friends were highly important to young people. Overall a high degree of resilience was apparent, supporting the need to move away from pathologising young people with a visible difference towards looking at how we can 'enhance resilience'.

Research consistently highlights the importance of social skills and interactions in mediating the effects of an altered appearance and in supporting positive adjustment and resilience for young people with a visible difference. The development of social and romantic relationships is a key developmental task for all young people and furthermore social skills may also impact on romantic relationships for young people with a visible difference. A recent study by Griffiths et al (2012) surveyed (online) 40 young people aged 13-20 with various visible differences. Seventeen participants felt their appearance prevented relationships developing. This group felt strongly that attractiveness is highly valued in romantic relationships, but they reported feeling unattractive and self conscious. However those who felt confident in romantic relationships (n=23) valued attributes other than appearance and also tended to normalize romantic anxieties, attributing them to a normal teenage experience rather than to their visible difference. Additionally those who were not anxious displayed confident social skills; a finding that further supports the notion that strong social skills support positive adjustment and management of a visible difference.

The importance of both friends and family is highlighted by Williamson et al (2010) who explored both young people's and parents' experiences of appearance change during cancer treatment. Data came from both case study interviews and an online survey. Twenty two young people (aged 13-18) took part along with six parents. In line with previous studies in this section, findings underline the importance of appearance to the young people in the study. Both young people and parents described looking different as being challenging. As reported by Wallace (2007, a & b) young people used strategies to appear 'normal' and to hide visible signs of illness. Williamson highlights the importance of managing the reactions of others; young people reported concerns relating to how others would react to their altered appearance. Social support was critical and family and friends were

able to provide reassurance and also acted as shields. As previously mentioned, peer shielding can be highly supportive and Williamson highlighted 'parental shielding' as a strategy that parents had employed. This involved intervening in social situations to deflect attention and defuse difficult situations, supporting friendships and being sensitive to their child's appearance changes and desires to manage these changes. Both parents and friends were found to play important roles in supporting the social interactions of the young people with a visible difference. Additionally, Williamson reports that young people wanted information about appearance changes during cancer treatment either from health professionals or families, and some would have liked opportunities to speak to experienced patients. Overall the family was found to support resilience and a more positive psychosocial outcome.

Also investigating how people 'live with' a visible difference, Prior and O'Dell (2009) interviewed four adolescents (aged 11-13) with different facial disfigurement and their mothers. Like Wallace's studies, the interviews were analysed using IPA. Findings highlight coping as an ongoing and changing process and friends again played an active role in facilitating social involvement. Bullying was a concern for the young people, particularly appearance-related teasing. This study further emphasises that adjustment is an active and evolving process both for adolescents and their mothers and findings again underline the importance of friends and family in adjusting to a visibly different appearance.

Both Williamson et al and Prior and O'Dell particularly highlight the central role that families play in young people's management of a visible difference. Families are important sources from which young people learn cultural norms about the salience of appearance and receive feedback about the way they look (Gillen & Lefkowitz, 2009). Wallace (2007 a & b) found that the value an individual placed on appearance before it was altered was found to be an important factor in resilience. Children receive direct messages about their appearance from their families as well as messages that are communicated less directly such as modelling behaviours and parents' negative comments about their own appearance (Bellew, 2012). The way in which families talk about, manage and support their children's appearance concerns are likely to be highly influential on young people and highly significant in positive adaptation to a visibly different appearance (Bellew, 2012). Indeed, Rumsey and Harcourt (2006) write that "*...family psychological processes are more important than biomedical variables in promoting adjustment*" (p117). Thompson and Kent (2001) suggest that parental emphasis on appearance in early childhood can result in 'appearance consciousness'; in the case of a child with a potentially stigmatising condition parents could unintentionally increase the emphasis on appearance through monitoring activity. This may be particularly relevant for young people with NF1 and their parents who may be aware that neurofibromas often first appear during puberty so may look out for changes to their appearance.

It is important to note that while there is a growing body of literature exploring young people's experiences of visible difference, this relies heavily on qualitative data and is often descriptive. In order to understand adaptation and consider the support and care needs of young people with a visible difference research needs to both explore and quantify how visible differences impact on children and young people. However, overall studies with young people with a visible difference

demonstrate that appearance is important to them, just as it is to most young people. This may mean that in addition to the usual changes to appearance that take place during adolescence, visible differences that have been apparent since birth take on a different meaning, due to developmental and social changes. As such adolescence is highlighted as a time during which young people's appearance concerns change, and a time during which interventions may be timely.

### **2.3 NF1: Focusing on young people and appearance**

The young people in the studies outlined in the previous section had a range of visible differences, situated on different parts of their bodies and resulting from different conditions and treatments, yet their concerns and accounts of challenges and positive adjustment are remarkably similar. However, these studies have concentrated on actual changes to appearance. A crucial factor for young people with NF1 is that those with few or no actual changes to their appearance may still have appearance-related concerns regarding worries over possible future changes.

The literature reviewed suggests that young people with a visibly different appearance may face social stigma. For those with NF1 this may be compounded by the genetic nature of the condition. It is also important to note that the nature of individual condition variables may be highly pertinent in adjusting to a visible difference. A recent meta analysis of body image of children and young people with chronic illness (Pinquart, 2013) concluded that young people with chronic illness had a less positive body image than those without chronic illness. Additionally differences between the body image of those with and without chronic illness was found to be illness-specific underlining the need for NF1 specific research that explores how young people with the condition manage and adjust to actual and possible changes to their appearance. Whilst issues around managing the appearance related aspects of NF1 have been mentioned throughout the existing literature and appear to be highly important within adult's accounts, no research to date has specifically investigated the role of appearance within the psychosocial adaptation and experience of young people with NF1. Similarly, whilst it is theorised that adolescence may be a challenging time for people with NF1 (particularly due to appearance changes and uncertainty) again this has not been directly examined.

Most of the research concerning the psychosocial impact of NF1 has concentrated on adults (eg Page et al, 2006; Ablon, 1999) or children (eg Wolkenstein, 2009; Oostenbrink et al, 2007; Barton & North, 2004). Much of the research reported as being with 'adolescents' has employed a young sample (for instance, each of the following studies claimed to focus on 'adolescence', but Graf et al's (2006) participants had a mean age of 11.6 years, Dilts et al's (1996) 10.5 years, Counterman's (1995) 11.8 years, Huijbregts et al's (2010) 12.3 years). The oldest mean age of participants in an adolescent study appears to be that of Sebold et al (2004) who report a mean age of 15 years. Interestingly they report differences between those under and over 15 years of age suggesting that young people's perceptions of their condition change during adolescence.

Many of the physical aspects of NF1 can change during puberty meaning that studies with a younger age group may have included a sample whose appearance has not yet been altered by the condition.

It is also important to consider that the psychological and social changes that occur during adolescence and early adulthood may alter the way in which young people understand NF1 and, furthermore, the increased awareness of appearance that is common during this life stage may impact on individuals' awareness and perception of their condition.

The importance of attending to young people's experience of NF1 is outlined by Oates et al (2013). The authors report that 15 of their 17 participants (Australian adults aged 25-33 with NF1) had not had a NF1 health check in many years, and most had not had a check since transition to adult healthcare. The authors describe that while these individuals had a typical range of NF1 symptoms at the time of transition, the following lack of care as a young adult was concerning and did appear to lead to greater numbers of NF1 related complications. It is unclear exactly why these young adults were disengaged from health care; however research has pointed to adolescence and early adulthood as a time during which individuals' experience of NF1 changes. While Sebold et al (2004) reported differences in perceptions of NF1 between participants aged over and under 15 years of age, Hummelvoll and Antonsen (2013) reported differences between their youngest participants (aged 18-25) and those aged 26-37 in terms of friendships, depressive difficulties and self confidence. Managing a chronic condition, psychosocial adaptation and the challenges associated with NF1 could be expected to differ throughout the life span, and in order to best support people throughout their lives it is important to examine how people's experience during adolescence informs the psychological impact and subjective experience of NF1.

Research consistently highlights the importance of individuals' social skills and social interactions in living with a visible difference. This finding may present a significant risk factor for young people with NF1; young people with the condition are reported as having fewer friends than peers, as well as difficulties with social skills and with social information processing. This could suggest that young people with NF1 may need support developing the skills needed to mediate the impact of an altered appearance. There appear to be major psychosocial risk factors for young people with NF1 related to possible or actual appearance changes alongside the usual social and emotional changes associated with adolescence. Appearance concerns are reported as a central concern for adults with NF1, regardless of clinical severity (Smith et al, 2013; Granstrom et al, 2012; Wolkenstein et al, 2001; Ablon, 1999). However there is a dearth of research that investigates the experience of young people with NF1 or focuses on the appearance related aspects of the condition. Furthermore, subjective and objective perceptions of severity and the impact of the condition may also be very different, between young people, medical professionals and family members. Sebold et al (2004) specifically examined the relationship between adolescents' families' perception of the severity of NF1 and the clinical severity. While parental and adolescent findings correlated, adolescents reported less of an impact than parents. A similar finding was reported by Counterman et al (1995) who again found parents and children rated the severity of NF1 symptoms similarly but they differed in their perceptions of the child's adjustment to this severity.

Adolescence and early adulthood is a crucial life stage for the development of adaptive skills and body image development and is highlighted as a particularly challenging time for people with NF1 (eg

Ferner et al, 2012; 2007; Wellman, 1990). Neurofibromas often first appear during puberty and not knowing how the condition will progress makes adolescence a time of greater uncertainty. Retrospective reports by adults with NF1 suggest adolescence may be a particularly difficult time, regardless of severity or noticeability of the condition (Wellman, 1990). In addition to managing actual changes to their appearance caused by NF1, difficulties for young people may stem from managing the possibility of appearance changes caused by the condition and from the unpredictability of the condition (Ferner et al, 2007; Ablon, 1999). It may also be important to learn about NF1 before adolescence, since growing up with the knowledge of having the condition may make acceptance and coping easier (Rubenstein & Korf, 1990).

In addition to describing and understanding the negative psychosocial experiences of people with NF1, it is important to understand why many people may feel positive. For instance, whilst 48% of adult participants in Benjamin's study (1993) reported problems related to appearance aspects of NF1 at school, 52% did not. It is important to ask what it is about this group that meant they did not report appearance related problems at school. Additionally one third of participants in Smith et al's study (2013) report their primary appearance concern was not NF1 related, a reminder that NF1 may not be an individual's main concern, young people with NF1 may be more concerned about other aspects of their appearance, or may feel positive and happy with the way they look.

By understanding the role of appearance within the day-to-day lives of young people with NF1, this thesis aims to explore both challenge and resilience. Through understanding positive adjustment we are better equipped to understand and support those who may need support. As Wellman (1990) writes "*the specific developmental patterns and needs of a teenager are not changed by NF. It helps to recognise that and to let the teen take on the world on his or her terms. NF is part not the whole, of the teenager's life*" (p214.)

## **2.4 Conclusion**

The literature reviewed in this chapter highlights that NF1 may have a profound impact on young people's overall experience in many diverse ways. In particular, psychological issues for young people with NF1 may relate to both actual and potential changes to appearance. Living with a chronic illness that may or does alter appearance may present many challenges for young people (Pinquart, 2013; Suris et al, 2004). Adolescence is potentially a transitional point in dealing with appearance concerns (Feragen & Borge, 2010). Young people with NF1 have to negotiate managing an unpredictable chronic condition which could alter, or is altering, their appearance during a time in their lives in which body image is often a particular emphasis. Young people with NF1 may also have learning and behavioural difficulties, in addition to impaired social skills, all of which may have a bearing on managing an altered appearance. For young people to have difficulties in this area could hinder positive adjustment, since social skills and interactions are critical in mediating the impact of a visible difference.

However, it is unclear from the current literature how young people manage an actual or possible altered appearance resulting from NF1, what influences positive adjustment and what support they may need. Recent studies with adults with NF1 highlight the impact of the condition on general appearance concerns, social self confidence and body image while questioning the impact of the visibility of the condition (Smith et al, 2013; Granstrom et al, 2012). Yet, while research with adults with NF1 suggests that appearance plays an important role within their experience of NF1, how and when this becomes important is unclear. While some studies have included young adults, there is no research that specifically examines young people's experience of NF1 during adolescence and none that concentrates specifically on managing actual or potential changes to appearance and how this impacts on young people. Understanding the particulars of young people's lives can support the development of services designed to support the positive adaptation of young people during adolescence and into adulthood. Gilboa et al (2010) suggest that future research should include personal input from those who have the condition. Furthermore they propose research should contribute to a better understanding of the experience of those with the condition in order to develop interventions focusing on making a difference to the lives of individuals with NF1 and their families.

Overall the literature outlined in this chapter particularly highlights the need for research that describes and examines the daily lived experience of those affected, focusing on how young people manage appearance changes and the possibility of changes to their appearance.

Specifically, the literature reviewed suggests that:

First, adolescence and early adulthood is an important developmental stage in terms of health and appearance, yet this may be a particularly difficult and challenging time for people with NF1 due to uncertainty, physical and social changes as well as a growing awareness of the condition. Studies which have included older adolescents or younger adults highlight this as a time of change. However few studies have focused primarily on this life stage meaning that there is not a clear picture of young people's experience of the condition.

Second, whilst appearance concerns are often cited by adults with NF1 and described by researchers, the nature of these concerns and how and when they may develop has not been specifically examined in young people with NF1. In order to support young people to become resilient adults it is critical that their relationship with their appearance is clearly understood.

## Chapter 3: Methods

The first two chapters of this thesis highlighted the importance of focusing on the issues associated with appearance for young people with NF1. This chapter considers methodology and how to explore these topics. The research aim is outlined, the approach of this thesis and a critical account of the use of different methods is provided. The chapter then considers how to ethically explore young people's experiences. This chapter serves to provide a broad overview of methods of investigation; more detailed descriptions of the methods used within individual studies can be found in Chapters 4 to 8.

### 3.1 Research Aim

This thesis aims to provide a comprehensive, in-depth understanding of the role that appearance plays within young people's experiences of living with NF1, in order to inform healthcare provision and the development of support.

This aim was addressed in two stages using a mixed methods approach (see figure 2 below). The first qualitative stage employed semi-structured interviews (Chapters 4, 5 and 6) to explore individual experience. The findings from this stage informed the development of questionnaires used in the second stage (see Chapters 7 & 8). Participants included young people with NF1, alongside parents of young people with NF1 and health professionals.

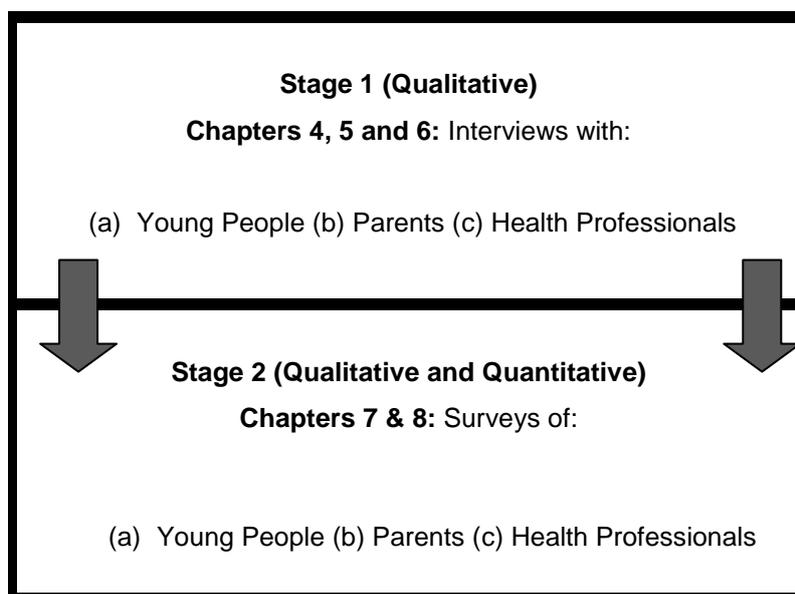


Figure 2: The Overall Research Plan

### 3.2 Methodological considerations

Willig (2001) suggests that in order to decide on what methods to employ researchers need firstly to consider the research question and their epistemological position. The starting point for considering methods of investigation in this research was a reflection on the research aims and a consideration of how experience can be communicated and investigated. Essentially this research adopts a pragmatic position, recognising that experience and knowledge are produced in different ways, accepting that there is valid information in personal subjective stories and in psychometric testing and statistical analysis. Creswell and Plano Clark (2011) suggest that within a pragmatic approach; *“The focus is on the consequences of the research, on the primary importance of the question asked rather than the methods and on the use of multiple methods of data collection to inform the problems under study”* (p41). A pragmatic approach means that methods are chosen by virtue of their practical value for the specific question being asked at a particular stage within a project. In this way the use of method is dependent on a range of factors rather than a guiding philosophy (Denscombe, 2008). Pragmatism allows researchers to *“...frame their work in terms of its intended consequences and what they hope it will achieve”* (Dures et al, 2010 p3). A pragmatic approach is particularly well suited to exploring health issues as it engages with the complexity of health and health care (O’Cathain et al, 2007).

By using both qualitative and quantitative methods it is possible to examine individual experience in rich detail and also investigate and measure the commonality of experiences presented. This research broadly explores experience through in-depth interviews and then focuses, quantifies and examines variables identified using questionnaires. The employment of both qualitative and quantitative methods categorises this research as having a mixed methods design (Brannen, 2005). A mixed methods approach draws on different methodologies and theories of knowledge production at different points in the research process, with the aim of them becoming entwined in the theoretical position or story behind the research as a whole. It is based on the assumption that there are multiple viewpoints and truths which can be accessed through both qualitative and quantitative research methodology.

Qualitative research is based on the perspectives of those being studied (Elliot et al, 1999) and as such it can be categorised as highly contextualised; researchers seek holistic in-depth accounts of individual experience based on the theoretical position that reality is subjective and constructed. A common criticism of qualitative methods is that they lack reliability; findings cannot be generalised and as such it can be difficult to draw conclusions (Castro et al, 2010). However qualitative methods can bring valuable insights into people’s experiences of health and illness and furthermore reliability may be an inappropriate construct when researchers are suggesting one possible interpretation, rather than generalising (Yardley, 2000). Qualitative methods are widely used in investigating adolescence and health; as Rich and Ginsburg, (1999) write *“Our vision of the direction of adolescent health is likely to be clearest when adolescents serve as our guides, for they best understand the prose and the poetry of their lives”* (p. 377). From this perspective qualitative methods can be seen as

empowering; participants are not necessarily constrained by the questions asked, they can elaborate and deviate providing their own rich descriptions of their lived experience.

While this thesis aims to be grounded in individual detail it also aims to explore commonality by comparing different accounts in order to develop an understanding of the common challenges young people with NF1 face as well as examples of resilience. Understanding in this area can then be used to inform the development of interventions and recommendations for supportive care. To this end, quantitative methods are also employed. Quantitative research allows for greater numbers of people to participate in research and have their voice included. It allows for commonality of experience to be viewed statistically and can give a strong indication of the importance of these experiences to those affected, furthermore it allows for testing of generalisability (Creswell, 2009). Quantitative approaches are based on the view that reality is universal and objective and the strength of a quantitative approach relates to measurement and comparison, and can test associations between variables. However it can lack ecological validity, as it can be removed from people's day to day lives.

The differences between qualitative and quantitative methods, in particular the way in which they draw on different assumptions about the nature of reality (ontology) and ways of understanding (epistemology) led to debates during the 1970s and 1980s regarding the superiority of each method, described as the '*paradigm wars*' (Creswell & Plano Clark, 2011; Tashakkori & Teddlie, 1998). A paradigm can be defined as a '*set of basic beliefs*' (Guba & Lincoln, 1994 p107). Quantitative methods are underlined by the positivist paradigm which views truths as real and discoverable while qualitative methods draw on the constructivist paradigm which views reality as socially constructed, believing that there are multiple truths. Essentially, qualitative research explores through an inductive and descriptive process of interpretation while quantitative research identifies associations and differences; predictions are made using hypotheses and theory. Qualitative elements explore while the quantitative measure and correlate (Dures et al, 2010), reflecting the way in which we often experience the world; through interpreting a mixture of different types of information.

During the late 1980's into the 1990's, the use of mixed methods went through a period of development during which procedures were mapped and defined (Creswell & Plano Clark, 2011). In recent times combining qualitative and quantitative methods and adopting a mixed methods approach has become widely accepted as a method in its own right (Bergman, 2008). It is recognised as a distinct third research approach or paradigm, in addition to qualitative and quantitative research (Creswell & Plano Clark, 2011; Johnson et al, 2007; Bryman, 2006). There is now a journal dedicated to mixed methods research and a series of textbooks available for researchers. Using mixed methods has become common in health research (Creswell et al, 2004) and has previously been used in appearance research with adults (Dures, 2012). For instance Dures et al (2011) describe how mixed methods were used to explore the psychosocial impact of the genetic skin condition epidermolysis bullosa (EB) on adults. A qualitative approach was employed in order to explore experience inductively while a second quantitative stage enabled an exploration of the prevalence of aspects identified in the qualitative stage, so while interviews highlighted aspects of individual appearance concern, surveys demonstrated statistically significant predictors of wellbeing. The use of different

methods of research and the integration of quantitative and qualitative approaches should allow for a more complete understanding of the research area than either method would alone.

Mixed methods research can adopt many different individual methods; Feilzer (2010) writes that while it is important that research is robust, transparent and easily replicated, "*Pragmatists do not 'care' which methods they use as long as the methods chosen have the potential for answering what it is one wants to know*" (p14). However use of this paradigm should not mean that methods are not given careful consideration, methods should be employed with an explicit purpose. O'Cathain et al (2008) point out that in their examination of mixed methods proposals and studies most did not explicitly describe and justify the use of mixed methods. In using mixed methods there is a need for researchers to provide a rationale that considers why using both qualitative and quantitative methods will support their research aims (Dures et al, 2010). With this in mind the following section stipulates how the use of mixed methods supports the overall aims of the thesis.

### **3.3 The use of mixed methods in this thesis**

Creswell et al (2003) suggest when conducting mixed methods research, researchers should specifically consider (1) the sequence of methods, (2) whether research is driven by a particular method and (3) how they are to be integrated. This section considers these three points in reference to how methods were employed in this thesis.

#### **(1) The sequence of methods**

The first stage of this research involved semi-structured interviews while the second stage employed questionnaires. The research began with broad individual accounts which were analysed alongside a review of pertinent literature, to inform the development of primarily quantitative surveys which were used to explore how common the experiences identified in the first stage are amongst a larger sample. In addition, relationships between different areas of experience within and between participant groups were investigated. This design can be categorised overall as a sequential, exploratory mixed methods design, as outlined in figure 3. A sequential design explores a topic in one stage building to a second phase in which initial findings are tested further and/or generalised (Creswell et al, 2008). In this thesis, it is important to note that the second stage is itself comprised of both qualitative and quantitative data, which is viewed as complimentary and merged and analysed concurrently.

Particular methodological issues within a sequential design relate to whether the same or different participants will be involved in both stages, as well as how many participants will take part. In this thesis the samples are not being compared but rather the second stage builds on the first. As the aim of this research is to explore more widely and generalise in the second stage, a greater number of participants were included at that point. In part this is also recognised as a pragmatic choice. Questionnaires are more widely accessible to greater numbers of participants than individual in depth interviews; they are also less time consuming and as such may be more appealing to participants for

this reason. However, a particular consideration when designing this research was that some participants in the second stage may want to spend time reflecting on their personal experiences and may feel that ticking boxes on a questionnaire does not fully represent their unique experience. In acknowledgement of this surveys included open qualitative questions so that participants were able to expand on issues raised, should they wish to. This approach aims to ensure that participants feel that they are being treated as an individual and have the opportunity to write about aspects of their experiences that they feel are important.

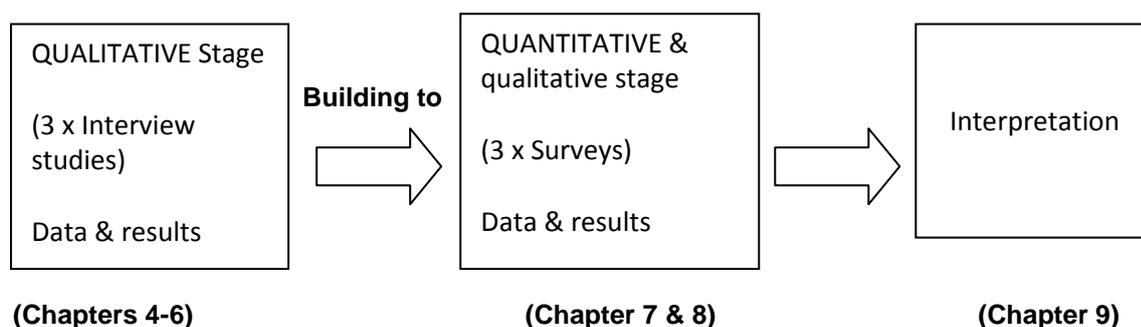


Figure 3: The Exploratory Research Design, as used in this thesis (capital letters indicate the more dominant methods)

While details of recruitment for each study can be found in individual chapters, it is useful to note at this point that recruitment for the first stage took place through support groups and contacts in the UK whilst participants for the surveys were recruited internationally through support groups, professional organisations and also through NHS clinic lists in the UK. This reflects pragmatic recruitment considerations as well as the nature of a smaller exploratory, in depth, interview stage and a broader survey.

All interviews were planned to be conducted in person and as such there were pragmatic constraints regarding how far the researcher could travel to conduct an interview. With this in mind it was decided that the interviews studies would be open to participants in the UK only. For the second stage, surveys were available both as paper copies and online. In particular, the online availability of the surveys meant that these were accessible to participants anywhere in the world, as long as they had access to the internet and could read and write in English. A particular consideration within this was whether the international nature of the survey would make findings less applied, as respondents would be reflecting on different health care systems. However, while many aspects of healthcare management may be different, this research concentrates on the role of appearance within young people's management of NF1 and this may be more universal.

The decision to recruit through support groups for the interview study and then through both support groups and NHS clinics for the survey stage related to the desire to interview a range of young people and parents, with mild and more complex NF1. NF1 specialist clinics are likely to see complex cases of NF1 and individuals on these lists may not be representative of many with the condition. In a large

survey, recruiting from many different avenues, this may not have a great impact on findings. However in a smaller exploratory interview study it was considered that this may have led to a less representative sample. Whether these decisions regarding recruitment impacted on findings is evaluated in Chapter 9.

## **(2) Whether research is driven by a particular method**

While some researchers have argued that a mixed methods approach can mean that qualitative research becomes secondary to quantitative (eg Denzin & Lincoln, 2005), others highlight that qualitative research can be highly prominent within a mixed methods research project, to the extent that a project can be qualitatively driven (Creswell et al, 2006). This thesis is driven by qualitative methodology which was the starting point of the research and was used to define the parameters of the survey. The dominance of qualitative methods reflects the view that a qualitatively-driven approach to mixing methods has great potential for exploring social and lived experiences (Mason, 2006). Chapters 1 and 2 highlighted the need to explore the impact and role of appearance for young people with NF1. As there is no known research that concentrates specifically on this, it is necessary that this research starts by defining the aspects of appearance that are important to individuals. Therefore in order to ensure this thesis is grounded in young people's actual experience, the starting point of the research was to talk to them, their parents and experts in the area (health professionals). In-depth interviews were chosen as this qualitative method is particularly well equipped to gain rich in-depth accounts of people's different and complex experiences (Rich & Ginsburg, 1999). Interviews were designed as semi structured allowing for new insights to be discovered rather than just focusing on predefined areas.

Although surveys in the second stage of the research employed mixed methods, these are primarily quantitative. Quantitative data was sought at this point to allow for a focused examination of specific variables enabling comparisons within and across data sets. Quantitative research focuses on deduction and confirmation, it tests theory, explains and predicts. It uses standardised data collection and analyses findings statistically providing an understanding of broader trends and exploration of specific variables within a population. The strengths of a quantitative approach include accurate measurement of a construct, the ability to make comparisons and to examine associations between variables and test hypotheses (Castro et al, 2010). Quantitative data is particularly useful in informing care and support provision, in order to plan interventions and care health professionals and support groups are likely to value quantitative data that not only describes a variable but also indicates its strength and prevalence.

## **(3) How methods are integrated**

Bryman's (2008) main concern with mixed methods research is that it is often insufficiently justified and in particular he also highlights the need to be explicit in detailing how methods are mixed. With this in mind, the process of mixing methods is outlined here and described in detail at each point of

integration. There are three points at which qualitative and quantitative data are integrated. First, the analysis of interviews provided the framework for the development of the survey. Second the qualitative and quantitative findings within the questionnaire data were analysed and reported simultaneously. The third point occurs in the final chapter of this thesis in which findings from across the thesis are synthesised.

The first point of integration was the analysis of interviews and the identification of areas to take forward in surveys. Analysis needed to be flexible so that it could be used within each interview study, exploring direct personal experience and more professional insights. It needed to capture the detail of participants' experience which could be expected to be highly variable, reflecting the variability of NF1. As such interviews were analysed using thematic analysis. Thematic analysis is a way of encoding qualitative data and aims to identify patterns across a dataset in order to answer a research question (Braun & Clarke, 2006). Thematic analysis is theoretically flexible, meaning that it can be used within a variety of frameworks, including the pragmatic approach taken within this thesis. It is particularly well suited to exploring individual experience (Boyatzis, 1998).

Themes are patterns found in the information, in this case interview transcripts, which describe, organise and interpret a phenomenon. Themes can be generated either inductively (i.e. are data driven) or deductively (i.e. are driven by theory) (Braun & Clarke, 2006). The thematic analysis employed in this research was inductive. An inductive approach allows for the generation of unanticipated insights (Boyatzis, 1998) and this was particularly well suited to the first stage of this research which was designed to be exploratory in nature and was not guided by any particular theoretical perspective. Thematic analysis aims to remain close to the data and is therefore grounded in the experience and words of those who participate in research. Yet, while remaining grounded in individual experience thematic analysis is able to summarise features of a dataset and highlight the similarities and differences within individual accounts (Braun & Clarke, 2006; Boyatzis, 1998). This was particularly relevant within this research as the aim was to explore individual accounts in order to determine areas to take forward and develop in a questionnaire.

Thematic analysis uses constant comparative analysis processes and each interview study was analysed using the six stages of thematic analysis as outlined by Braun and Clarke (2006). Each study (1-young people, 2-parents and 3-health professionals) was analysed separately and a different notebook was kept for each analysis. This notebook contained reflexive notes from the interview stages and further thoughts were added during the first stage in each analysis, which was to transcribe interviews and then read through transcripts. Transcripts were read several times in order to become familiar with the data set. Following this, the next stage was to generate initial codes. This led to considering which aspects of the dataset could be coded. Codes are the most basic aspect of the data and describe features of the data. Coding is an important aspect of analysis (Miles & Huberman, 1994) and transcripts were systematically coded line by line. This was done manually using highlighter pens and writing notes on the transcripts. Codes at this stage were both descriptive and analytical. Some codes simply described what the participant had said for instance if the

participant discussed school generally this was coded as 'school' however if they talked of difficulties at school this would be classified as 'school' then in brackets (negative) adding a layer of detail to the code. Codes were clarified and defined as this stage took place and once all transcripts had been coded they were read again and examined to ensure codes were consistently applied. However, as accounts were diverse it is important to note that a deliberate attempt was made to ensure that less common aspects were still coded and variety was not lost.

Following coding, the next step involved exploring if and how the initial codes could be grouped into themes across each data set. This involved reading the reflexive notebooks and writing up a full list of codes that had been applied. These codes were reviewed and collated into clusters. This entailed moving back and forth between the list of codes and data extracts. Groups of codes were defined as a potential theme when they appeared to fall under a common definition and did not overlap with another group of codes. At this point draft thematic maps were produced and working titles were given to each theme.

The fourth phase (Braun & Clarke, 2006) involved reviewing themes. This process involved moving backwards and forwards between proposed themes, initial codes and data extracts. Draft themes were considered and some were broken down into separate discrete themes, others were combined with themes that seemed similar. Themes were then considered in light of each data set as a whole asking whether they accurately described and captured the original data. Following this stage, the fifth phase was to critically re-examine themes in order to produce clear definitions, and identify subthemes. This process produced a final thematic map which illustrated the themes within each analysis diagrammatically (see figures 4, 6, 8 in Chapters 4, 5 & 6). At this point each theme was given a clear definition and a description of each theme and subtheme was written. Braun and Clarke define the sixth stage as writing the report. In this case the sixth stage can be considered Chapters 4, 5 and 6 which describe and analyse the interview data using examples from the data.

At all points during the analysis, codes, themes and sub themes were discussed within supervisory meetings. Any differences in opinion were discussed until a consensus was agreed. In order to ensure rigour and quality of the research during analysis Yardley's (2000) criteria for assessment of qualitative research were used. An evaluation of the qualitative stage in relation to these guidelines is presented in Chapter 9 (section 9.4).

The analytic process generated a detailed and rich description of each dataset which led to the identification of the key aspects of young people's experience of NF1 from the perspectives of young people, parents and health professionals (figures 5, 7, 9 in Chapters 4, 5 & 6). These key aspects informed the development of a series of research questions, (outlined in Chapter 7) to pursue in surveys exploring the role of appearance within young people's experience of NF1. Literature was examined in order to identify existing psychological measures for use in surveys of parents and young people and both quantifiable questions (derived from quotes from interviews) and open ended questions were developed for use in all three surveys. This use of data from the interview studies to

define key aspects of experience, research questions and develop surveys is the first point at which methods are integrated and Chapter 7 clearly details this process.

The aim of the second stage of this research was to investigate specific aspects of the psychosocial impact of NF1 in a narrower and more defined way, thus allowing for generalisability of findings that originally derived from individual personal experience. The analysis of surveys is described in Chapter 8, quantitative data was analysed using a statistical software program (SPSS) and qualitative data was analysed using content analysis of open ended questions. The different types of data were analysed and reported concurrently in answer to the specific research questions outlined at the start of Chapter 7, therefore this was the second point at which methods were mixed.

The third point at which methods were integrated is in the final chapter of this thesis (Chapter 9) which discusses findings from across the thesis, and considers the impact of appearance on young people with NF1 and how findings could be applied in order to support young people. At this point the methods themselves, and the use of mixed methods is also evaluated.

### **3.4 Researching young people's experience**

Having discussed the theoretical use of mixed methods and then outlined how methods are used and integrated within this programme of research, this section considers ethical and practical factors involved in researching young people's experiences. In particular, who to include in the research and how to address possible power imbalances between young people and researchers is discussed.

Young people, parents and health professionals were included in both stages of this research with the aim of obtaining a holistic picture of the role appearance plays within young people's experience of NF1 from multiple perspectives. These three groups provide an individual and personal perspective, an understanding of the immediate family environment and an understanding of wider issues around the role of appearance within health care and management of NF1. In addition they combine to form a portrait of experience during adolescence into adulthood. Findings from the three groups are compared and discussed in order to explore and highlight different viewpoints.

The young people included in this research were aged 14-24 years old in mid adolescence and early adulthood. During this time, young people transition into adulthood; as highlighted in previous chapters it is a time of great psychosocial change as well as structural change as they move into adult healthcare and become increasingly independent. Researching young people's experiences of a genetic condition and how they feel about their appearance could be considered a particularly sensitive subject area. As discussed in the introductory chapter, it is important to include young people in research, however it is also important to acknowledge that this inclusion necessitates considering and addressing the particular ethical implications of undertaking research with minors. Particular considerations include first ensuring that young people and parents where appropriate, are fully informed about the research so that they are able to provide informed consent and participate in

the research in a meaningful way. Following this a second consideration is how to conduct research in order to facilitate the meaningful participation of children and young people.

With regards to informed consent, as noted in Chapter one British law is not clear cut with regards to the age at which children and young people can provide informed consent for medical treatment. Again with regards to consent to participate in research there is a great deal of ambiguity and debate. The Royal College of Pediatrics and Child Health (RCPCH) suggest that parental consent should be sought for young people under the age of 18. However the British Psychological Society code of human research ethics ([www.bps.org.uk](http://www.bps.org.uk)) stipulates that parental consent should be sought for young people under the age of 16. Rather than prescribing an age, the UK National Research Ethics Service (NRES) stipulates that the Gillick principle can be applied to consent to research meaning that young people can take part in research depending on their competency to understand the research (National Research Ethics Service, 2009). The test of Gillick competence refers to a case brought to the UK House of Lords which determined that children under 16 do not lack the capacity to consent due to their age but rather individual competence understanding and intelligence (Parekh, 2007). While assessing individual competence is important this can be highly ambiguous therefore for the purpose of this research a decision was made to ask all young people aged 14-24 to consent to take part in the research and to also ask for parental consent for young people aged under 18 years in the qualitative stage and under 16 years old in the survey stage. The process of how consent was sought is described below and in detail in later chapters however it is important to note that the difference in ages of consent between the two stages reflects the view that parental concerns may be heightened in qualitative research (Schelbe et al, 2014).

In order to ensure that all participants had sufficient information in order to provide informed consent information sheets and letters were sent to potential participants prior to interviews explaining why the study was taking place and what topics, in general terms, would be covered. Once young people contacted the researcher to arrange an interview further details were provided either orally or in email format. Before interviews began the researcher again discussed the aims of the research and assured participants that they were free to pass on any questions they were unsure of and could stop the interview at any point, It was at this point that young people, and a parent if the participant was under 18 years, were asked to sign consent forms.

It was recognised that some young people (both over and under 18 years) may wish to have their parent present during interviews. In order to facilitate this a decision was made to explicitly ask young people at the time of arranging interviews to consider whether they would like a parent present and young people were encouraged to talk to their parent/s about this issue. Before interviews commenced all young people were asked if they would like a parent present.

During the survey stage if young people were aged 14 or 15 their parents were asked to sign a paper copy of the survey in order to provide consent, or in the case of online surveys young people were asked to provide a parent's email address and parents were asked to confirm they were happy for their child to participate. In order to ensure that young people and parents were able to make an

informed decision letters and information sheets outlining the research aims and plans were sent physically with paper surveys and were also part of the online process.

In addition to obtaining informed consent a particular consideration was that the variability of NF1, and the age of participants meant that it was possible that some may not have thorough knowledge and understanding of the condition. As such, interview guides and questionnaires were worded so that participants were not asked directly about issues such as future prognosis or transmissibility/reproductive decision making. Instead these aspects were discussed if the subject was mentioned by the participant themselves and open ended questions and text boxes were provided on questionnaires so that, should participants wish to give additional detail, then they had the opportunity. While this may have meant some questions were unasked, and thus unanswered, it was felt this was the most ethical way to ensure the research process did not introduce areas of concern to young people.

Another consideration was whether to ask participants about specific learning and behavioural difficulties. As outlined in previous chapters many young people with NF1 may have associated learning and behavioural difficulties, including ADHD and ASD. However, previous research (see section 2.1.1) also suggests that children with NF1 may have specific difficulties with attention and processing social information without meeting the criteria for ASD or ADHD (Isenberg et al 2013; Huijbregts, et al 2010a, 2010b). In addition, some young people in this research may have had undiagnosed learning and behavioural difficulties. The manner in which learning difficulties are recognised, diagnosed and impact on individuals can differ greatly and asking and recording known instances may not reflect the intricacies of individual experience. Furthermore, an important aim of the interview study in particular was that participants were able to describe the aspects of their experiences that were important to them. Whilst having a diagnosis of a learning or behavioural difficulty may be highly relevant to one person, it may feel inconsequential to another. For the purpose of the interview studies a decision was made not to record or explicitly ask about learning difficulties but instead to ask about school in general terms. If participants mentioned learning difficulties then these were discussed. This decision was then revisited during analysis of interviews and the development of questionnaires.

An important tenet of this thesis was not to assume that young people would have appearance concerns, or that there was a relationship between the severity of visibly different appearances and feelings about appearance. The literature reviewed in Chapter 2 highlights that evaluating who may or may not have appearance concerns is highly complex and individual. Whilst some young people with NF1 may be very concerned about their appearance and the possibility of appearance changes, others may not. Furthermore, some participants may not be aware of the different ways in which their appearance could be altered by different symptoms of NF1. Developing an interview guide about young people's experiences more generally, rather than only about appearance, was viewed as a sensitive way of approaching interviews. This also reflected the findings reported previously from studies with young people with a visibly different appearance in that the way in which appearance

impacts on young people is complex and multifaceted. The aim of interviews was to ask young people about their experiences across childhood and adolescence (see interview guide, appendix 7). As interviews progressed and experiences were discussed, the interviewer focused in and asked questions that explored areas identified in the literature reviewed in the previous chapters about aspects of appearance which may be important to young people such as meeting new people or starting secondary school. This process enabled a discussion of appearance within the context of overall experience and the interviewer's questions and later analysis focused in on specific issues.

Having decided that interviews should explore the role of appearance within young people's experience, whilst not introducing areas that may be unknown to participants and not assuming that learning difficulties and appearance are 'problems', the next step was to consider how to explore experience ethically. It seems clear that in order to understand a young person's perspective and encourage participation they need the space and opportunity to express themselves.

Participants in this research were aged from 14 years upwards. A research interview can be daunting for anyone and for a young person this may be especially so. Researchers need to think about how young people communicate and find a way into discussing and exploring experience. For instance, young people may be used to adults asking questions that have definitive answers and methods of research need to address these issues so that young people can fully participate.

Literature related to research with children and young people was therefore reviewed. A common criticism in the field of children's research is that it has been largely about them rather than involving them (Hill 1997). Kirk (2007) suggests that the reasons for not including children in research generally relate to concerns that data from children may be unreliable and that they are a vulnerable group. However both of these points have been disputed (Mauthner & Doucet 2003).

The view that researchers have of children and childhood has changed a great deal over time. Previous dominant theories of childhood, including socialisation theory and developmental psychology saw children as incomplete and or passive receptors of a socialisation process. These assumptions were questioned in the late 1980's leading to a greater interest in how children experienced their lives (Mauthner & Doucet 2003). A particularly dominant perspective at this time was social constructionism (James and Prout 1997) which sees children as active participants in their lives, and therefore possible of being active participants in research. However this shift in the conceptualisation of childhood does not mean that children should be treated in the same way as adults. It is essential that research with children adopts appropriate methods for engaging with participants.

Different ways of doing this have been suggested. For instance Davies (1998) highlights the importance of reflexivity, he suggests that when researching young people's experiences researchers need to find ways of ensuring that their own views and interpretations of childhood do not dominate. He also underlines the importance of not viewing children as one homogenous group. This view challenges researchers to question their assumptions of childhood. Mandell (1991) suggests that the

views we have of childhood are likely to guide how we approach research with children. She suggests adopting the 'least adult role' and aiming to act more like a child within the research process. However this is problematic, first this may be off putting for children and young people, and furthermore recognising that children are different but still hold valid experiences and viewpoints may be more realistic given the ingrained social and power differences between adults and children (James et al 1998).

Addressing this power imbalance and allowing children's voices to be heard may be the most importance consideration when researching children's experiences. With this in mind literature regarding participatory approaches was reviewed. Participatory research involves knowing that your actions are given attention and acted upon, which is empowering (Boyden & Ennew, 1997). Participation means children and young people are able to express themselves without adult constructs crowding them out. Boyden and Ennew suggest that this should involve less dependency on words. They suggest that visual methods can be used as a prompt or stimulus and the respondent can then give their own interpretation and explanation. Visual methods are an entrance point, a way of communicating and bridging the gap between participant and researcher; they break the ice and stimulate conversations that are respondent led. Visual research can advocate a collaborative approach. Prosser and Loxley (2008) explain that there is a continuum as to how participation and visual methods can facilitate involvement and discusses how visual methods can extend participation by giving participants a sense of ownership and empowerment. They also point out that people make sense of their lives through embodied experience, through the interplay of sensory relations which may not be accessible in numbers or words. Visual methods allow for increased participation which can be seen as a form of empowerment. Therefore visual methods have the ability to address the power imbalances that can be inherent in research with young people whilst giving them a voice in research.

Pink (2001: p.4) stresses that "*specific uses {of visual methods} should be creatively developed within individual projects*". The aim of interviews with young people was to explore their experience of NF1, concentrating on the role of appearance. With this aim in mind literature was reviewed concentrating on visual methods that have been used to explore sensitive aspects of experience throughout childhood and adolescence. Literature on adoption/fostering and in social work suggests that visual methods can be used in understanding life history (Meese, 2010). Life story work was first developed in the 1980s and is now an established part of social work practice with looked after children (Baynes, 2008; Ryan & Walker, 2003). It provides a structure for talking and discussing previous events in a young person's life, and can be used in challenging and emotional situations to explore the experiences of young people who are looked after or in care. This can be led by a professional, as a method of exploring life events that young people do not remember, or can be a joint exploration of events of significance throughout childhood. The process can result in books or videos however it does not have to have a result, the process of compiling a life story itself is beneficial, giving

individuals a structured and understandable way of talking about themselves. The use of recall is important in participatory research (Young & Barrett, 2001).

This idea of experience as a life story has been used in this research within the interviews with young people. Participants were invited to complete a visual timeline of their life history; this entailed drawing a line on a piece of paper with their date of birth at one end of the line and the current date at the other, and then filling in significant dates and events in relation to having NF1. This included medical information, schools and any other meaningful events. This approach reflects the researcher's belief that it is important not to assume certain events have more significance than others but rather to allow young people themselves to talk about the significant events and aspects of their life. Timelines have been used previously with young people in life story work (Boyden & Ennew, 1997). Timelines were used with young people to encourage engagement in the interview and to promote discussion, timelines themselves were not designed to be analysed.

While young people's direct personal experience is central to this research, a decision was made to also include parents and health professionals in the research. Parents are included as they are influential in young people's adaptation to a chronic illness and the development of body image (see sections 1.4 and 2.2) and as such their views and experience are highly pertinent to understanding young people's experience. Within this thesis the term parent is used to refer to biological parents and other carers who assume parental responsibility for a young person. Parents were asked to reflect both on their child's experiences and their own experience of parenting a child/young person with NF1.

The inclusion of health professionals reflects their importance in the management of care and support for young people. Furthermore health professionals are likely to work with a range of people with NF1, including children, young people, adults and parents of people with NF1. Health professionals are in a position to be able to consider the experiences of many different young people with the condition. As discussed previously (see section 1.5) health professionals hold an important role in relaying information and in supporting a young person (and their parents) across different ages and varying stages of understanding and managing a chronic health condition and visible difference. Interview guides asked parents and health professionals broadly about their observations and opinions of the impact of NF1 on young people (see interview guides appendix 8 & 9) and the second research stage focused more specifically on the role of appearance within this, through surveys.

An important aim of the first research stage was to explore experience broadly and, as outlined in this chapter, interview guides were semi structured and steps were taken to put participants at ease so that they could describe their experiences and opinions. However, it is important to acknowledge that the topics explored and questions asked during interviews were framed by the literature reviewed by the researcher outlined in Chapter 2.

By including the three participant groups in both interviews and the subsequent survey study, this research is grounded in the in-depth accounts of young people, parents and health professionals. Surveys then allow reports to be explored and quantified within and between participant groups, exploring similarities and differences. It is hoped that this understanding can then be used to inform future care and support provision.

### **3.5 Conclusion**

The aim of this thesis is to explore and describe the role that appearance plays for young people with NF1, in order to provide a basis on which to inform support and healthcare. The parameters, or topics researched within this, are defined through in-depth interviews and qualified by surveys of young people, parents and health professionals. The inclusion of the three participant groups and the use of both quantitative and qualitative methods allowed for the research to be driven by in-depth accounts of personal experience and opinion, whilst allowing for quantification, leading to a rich, detailed and comprehensive account of experience. By understanding young people's experiences and the opinions and experiences of their parents and health professionals involved in their care, this thesis aims to provide ecologically valid accounts of how appearance impacts on young people. How this understanding can be used to develop support and care for young people as well as the implications for health professionals and parents is discussed in Chapter 9.

## **Chapter 4: Interviews with young people**

This chapter details exploratory interviews undertaken with young people living with NF1. It describes the study design and interview process and presents analysis of qualitative data.

### **4.1 Aims**

There were two aims of this study:

1. To obtain a comprehensive, in-depth understanding of the role that appearance plays within the young people's experiences of living with NF1
2. To use this understanding to identify areas to be investigated in a subsequent survey

### **4.2 Method**

#### **4.2.1 Design**

The rationale for taking an exploratory qualitative approach is discussed in detail in the previous chapter. The research design primarily reflects the desire to ensure young people were empowered and heard from directly. A qualitative approach was chosen as a way of gaining rich in-depth accounts of young people's different and complex experiences (Rich & Ginsburg, 1999).

#### **4.2.2 Ethical Approval**

The study was approved by the Research Ethics Committee of the Faculty of Health and Life Sciences at The University of the West of England, Bristol.

#### **4.2.3 Recruitment**

For this exploratory study a decision was taken to recruit participants from the general NF1 population rather than through NHS clinics directly as discussed in the previous chapter. The subsequent survey (see Chapter 7) included recruitment through NHS clinics.

Recruitment took place with the support of The Neuro Foundation ([www.nfauk.org](http://www.nfauk.org), a UK support group for people with NF1 and their families). Advertisements about the research were placed on the support group's website and in its newsletter (appendix 1 & 2). Details were also available on the websites of Changing Faces ([www.changingfaces.org.uk](http://www.changingfaces.org.uk), a charity offering support for people living with disfigurement) and the Centre for Appearance Research. Additionally advertisements were posted online, on sites related to NF1 (see appendix 1).

The inclusion criteria were young people aged 14-24 years (inclusive) with a diagnosis of NF1 who were sufficiently fluent in English and capable of taking part in an interview conducted in English and lived in the UK. Young people aged under 18 years were asked to provide parental consent in order to take part in the interview. In line with qualitative principles the aim was to interview young people until saturation point was reached (when the collection of new data is not producing new insights or findings). In total 21 young people contacted the researcher. Twelve were later unable to take part; of these, four aged 14 and 15 years emailed the researcher and asked to take part but parental consent was not given. In three cases young people informed the researcher that their parents did not want them to take part after having read through the study information. These young people were thanked for their interest and informed that they would not be able to take part on this occasion without parental consent. The remaining young person did not respond to emails regarding the study after having read through the study information. Two people contacted the researcher but were not living in the UK. As discussed in the previous chapter (see section 3.3) it was decided that recruitment for interview studies would take place in the UK and the subsequent questionnaire study would be open to international participants. Finally six individuals who did fit the inclusion criteria asked for further information but did not make any further contact. In each case a follow up email was sent but no response was made. As such it is unclear as to exactly why they chose not to take part. Of the 21 people who contacted the researcher nine took part in interviews.

#### 4.2.4 Participants

Four of the nine participants were recruited directly from the advert placed in a Neuro Foundation newsletter, three through The Neuro Foundation website and facebook page, one through the Changing Faces young people's online discussion section and one through a Channel 4 discussion forum related to NF1 following its inclusion in a television programme called 'Embarrassing Bodies'. Parental consent was sought and given for participants under 18 years. (See table 1 for participant details; please note that pseudonyms are used throughout).

Pseudonym	Gender	Age	Parent Present at interview?	NF1 Inheritance Pattern	Highest educational level to date
Tina	Female	21	No	Unsure	University
Sarah	Female	23	No	New	Postgraduate
Joanna	Female	17	Yes, Both	New	College
Robert	Male	20	Yes, Father	New	College
Ros	Female	24	No	Inherited	University
Daniel	Male	24	No	Inherited	Postgraduate
Katie	Female	14	Yes, Mother	Inherited	School
Lucy	Female	16	Yes, Mother	Inherited	School
Mark	Male	21	No	New	University

Table 1: Interviews with young people: Participant demographic details

The mean age of participants was 20 years. As can be seen in table 1, inheritance was evenly divided between new or inherited, which is in line with reported NF1 patterns of inheritance (Ferner et al 2007). Some participants reported receiving very little or no treatment for their condition while others reported having many operations and procedures. Participants reported being medically affected by NF1 in many different ways and were seen by health professionals including NF1 specialist consultants, paediatricians, GP, geneticists and orthopaedics, one person reported not seeing any health professionals. Although they were not asked directly, participants reported a range of learning difficulties and behavioural problems associated with NF1 including dyslexia (learning difficulty related to reading and comprehension), ADHD (Attention Deficit Hyperactivity Disorder), ASD (Autistic Spectrum Disorders), Dyscalculia (a numeracy learning difficulty) and Dyspraxia (developmental co-ordination disorder, affects movement and co-ordination). Several said they had not been officially diagnosed with learning difficulties but had been told they probably had one.

#### **4.2.5 Interview guide**

An interview guide was developed through the literature reviewed in Chapter 2. As discussed in the previous chapter, experience was discussed broadly, although the focus was the role of appearance. Participants were asked what they understood NF1 to be and were asked about when they first knew they had the condition, how family and friends reacted, their medical experience, how NF1 impacts on their life on a day-to-day basis and specific questions about appearance. The guide can be found in appendix 7. The exploratory nature of the study and the desire to put people at ease meant that participants were encouraged to take a lead in the interview and tell their story in their words and in their own way and time.

#### **4.2.6 Procedure**

People who were interested in taking part contacted the researcher and were sent an information pack, including parental information if they were under 18 years (see appendix 3 & 4). Those who decided to participate contacted the researcher to arrange an interview. Participants in this study were offered the opportunity to use timelines during the interview. Using a timeline entailed drawing a line on a piece of paper with their date of birth at one end of the line and the current date at the other, and then filling in significant dates and events in relation to having NF1 between the two (see discussion in previous chapter and appendix 6). However it was also noted that some young people may not want to draw a timeline. As such, people were sent details about the option to use a timeline in advance and their utilisation varied, three participants chose to use timelines. Timelines themselves were not designed to be analysed but were used as an empowering way for young people to share their experience and facilitate discussion.

Eight interviews were conducted face-to-face and one by telephone (Ros), at the participant's choosing. Participants were located in the South/South West of England (n=2), Midlands (n=2),

London and Kent (n=2), Northern England (n=1), Wales (n=1), and Northern Ireland (n=1). Face-to-face interviews took place in participants' homes (n= 6), or another setting of their choice (n= 2).

As shown in table 1 several participants, including all those under 18 years, chose to have a parent with them during their interview. In some of these cases parents came in and out of the room and their level of involvement in the interview differed between participants and throughout the duration of the interview. At the beginning of the interview participants were given the study information sheet again (appendix 3). The research plans and rationale were discussed and any questions answered. Confidentiality was assured and consent forms (appendix 10 & 11) were discussed and signed by the participant, and a parent if the participant was under 18 years. Interviews lasted between 30 and 80 minutes and were audio recorded with the participant's express permission.

Participants were reminded that interviews could be stopped or paused at any time and that they did not have to answer any questions if they felt uncomfortable or uncertain of the answer. If participants were noticeably upset at any stage during their interview they were asked if they would like to pause or stop. Three participants did become quite upset but chose to continue. All participants were reminded of sources of support and were given details of avenues of support verbally and in writing. Once data was analysed feedback was sent to all participants along with a copy of the young person's survey for feedback.

#### **4.2.7 Data analysis**

Interviews were audio recorded and transcribed verbatim and detailed field notes were kept by the researcher after each interview; these were subsequently used during analysis to add detail to data and to reflect on observations. Since NF1 is a highly variable condition and participants could be expected to have a multitude of different experiences, thematic analysis was chosen for this study as it maintains the richness of data whilst allowing areas of commonality to be analysed. All transcriptions were coded and codes were developed into subthemes and themes, following the six stages recommended by Braun and Clarke (2006) as discussed in the previous chapter (see appendix 15).

### **4.3 Findings**

#### **4.3.1 Themes**

Central to accounts of their experience of NF1 during adolescence were the variability, unpredictability and visibility of the condition. Its effect on appearance was discussed in terms of adapting to a changing appearance and managing interactions with others. Analysis revealed three key themes, each with subthemes as shown in figure 6.

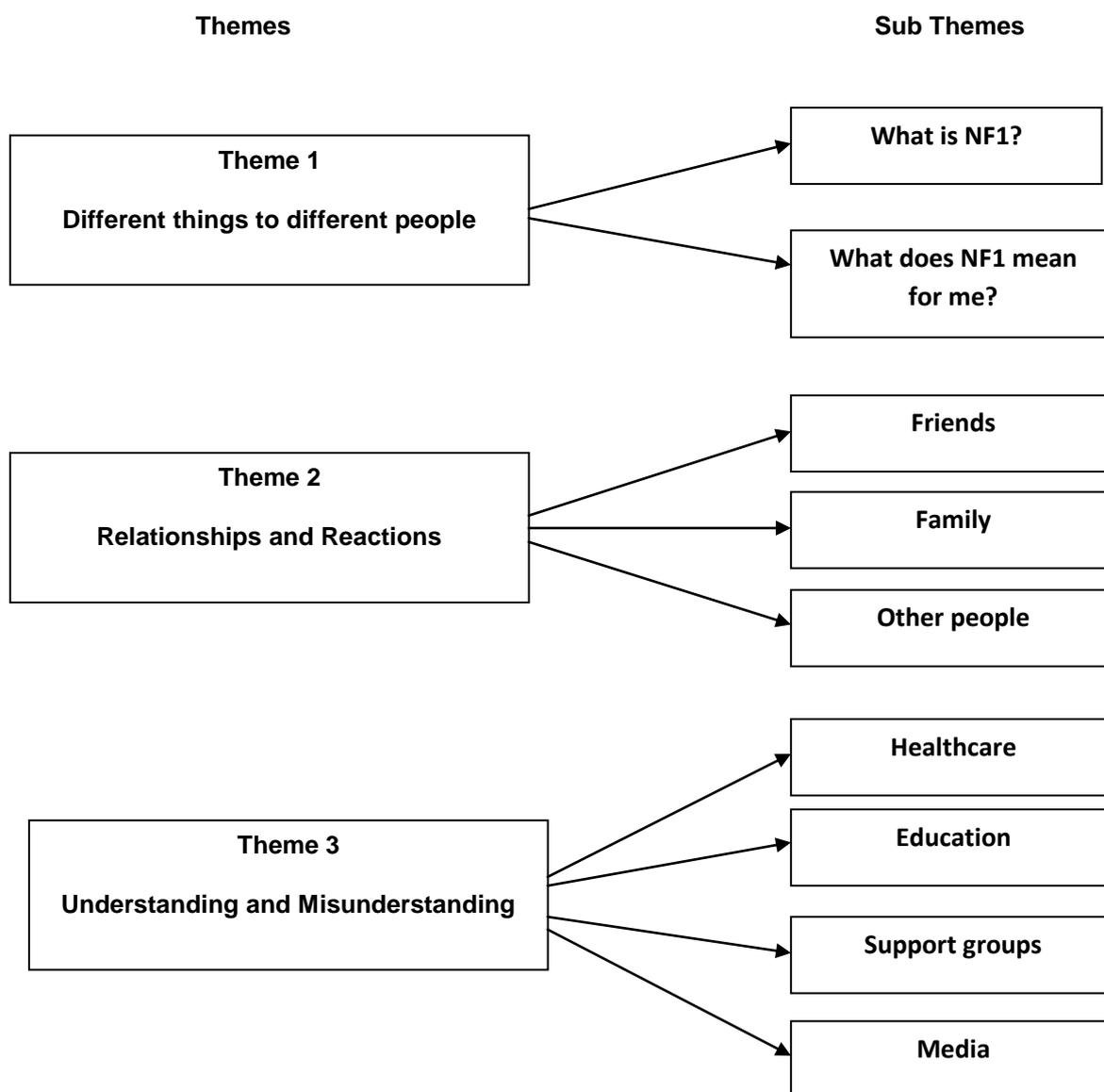


Figure 4: Thematic map of findings from interviews with young people

#### 4.3.2 Theme 1: Different things to different people

NF1 can be characterised by its variability and unpredictability (Ferner, 2007; Ablon, 1999). This was evident in two respects, firstly in how young people understood and conceptualised what NF1 meant to them and secondly in how they managed their own experience of a very individual condition. This theme is discussed within two subthemes of 'What is NF1?' and 'What does NF1 mean for me?'

##### What is NF1?

The variability of NF1 was particularly evident when young people described the condition. Some described it as a skin condition, some talked of 'tumors' and 'lumps', some referred to a specific part

of their body (eg legs), and others discussed learning difficulties or ADHD. As Daniel (24) commented: *"It's like a bag of pick and mix you don't know what you're going to get really, it could be anything"*

For most young people NF1 was just always part of their life, *'when you're growing up, like with every child, it's sort of.....you see it as normal, you don't question it.... I guess I just gradually knew more, more about it'* (Mark, 21). Others found out about NF1 later. Here, Tina (21) described her shock at learning she had the condition when she was 17 *"I knew there was something because I was finding lumps and stuff like that, I went to the doctors and they said like some other stuff but then like when they read my file they said 'hang on its neurofibromatosis' what the hell is that, you know what I mean?"*

While participants had varied experiences and symptoms, many felt their understanding of and feelings about NF1 had changed during adolescence. Katie (14), the youngest participant, described a growing awareness of having NF1 in recent years *'I don't know if I knew when I was younger or not and I have certainly become more self conscious of it'*.

For some this led to a desire to know more about the condition. For some this stemmed from physical changes; *"I think it was sort of early years of secondary school when I started to notice a few more of lumps and just a bit more curious about it and see how it was affecting me, and I yeah so I got a few more like information packs on it and stuff"* (Mark, 21).

Some felt that their conceptualisation of the condition altered as they learned more about NF1; *"I think when I was younger it was just the leg stuff that used to bother me, now it is more like the neurofibromas and stuff..... it had always been a leg thing and then to kind of reconceptualise that, it was quite weird .....it was just I've got NF that means my legs and my shin bones don't work properly so ....there's been a lot of learning since then"* (Sarah, 23).

### **What does NF1 mean for me?**

Accounts also varied regarding the emotional impact of NF1. Some participants felt it held little significance and that there was no real emotional impact at all, whilst others felt it had an extremely profound impact on their life. For instance Robert (20), commented that NF1 *"Bothers me now and again but it doesn't put me too down"* whereas Ros (24) explained that *".....if I didn't have NF I wouldn't have the problems I have got"*.

Participants described how managing the impact of NF1 involves dealing with the unpredictability of the condition. As Daniel (24) explained *"I may wake up next week with a tumour I don't know"*. People found different ways of managing the uncertainty, some tried not to think about it; Mark (21) said *"the more you think about it, the more it will affect you, so if you just thought yeah its fine, it doesn't matter"*

*then yeah*". Others felt it was important to be positive *"just be yourself and something like that and different people is what makes the world special"* (Katie, 14).

Demonstrating finding the positive, and also indicating that NF1 is stopping him from doing all that he would like to, Daniel (24) talked about the possibility of his leg being amputated should it break again and how he manages the uncertainty *"...I am so worried about breaking this leg because there is always doubt in the back of my mind if I do break my leg again the chances are they will get it amputated.....So you kind of... yeah that's the way I try to see the positive of it really I mean fair enough it would be I think rehab for a year or so then after that I would get a new stronger leg that may allow me to run, may allow me to play football or something it may allow me to do quite an active sport..."*

Sarah (23) explained how important it is not to assume the worst and to remain optimistic, *"I could live my life and have fewer problems than someone who right now is healthy but then might develop like diabetes or heart disease or something you know so there isn't, just because I've got NF now it doesn't really mean it doesn't necessarily mean that everything's going to go horribly wrong for me like when I'm older"*

Participants described finding aspects of NF1 challenging and distressing and discussed their concerns. Some felt NF1 impacted across all aspects of their lives and was the cause of considerable distress. For many, the impact of NF1 was related to its effect on their appearance. Participants discussed the visibility of NF1 in different ways, some referred to specific issues such as tumors, plexiforms or café au lait marks whilst others described the impact on the way they walked or the need for an aid such as a wheelchair, splint or orthopedic shoe. For many, visible symptoms and aids were a reminder of the condition and also signified to others that they had a medical condition.

Some were very distressed about the effect of NF1 on their appearance and the reaction of others *"I hated myself because I found out all these little marks and stuff little lumps...and I get frustrated by it because you know when it's like summertime and you want to be going out and go on the beach and what not and people see all these freckles on your body"* (Tina, 21). In talking about her appearance Joanna (17) explained *"I don't look in mirrors that much 'cos it's like really big when I look in mirrors. The cheek looks massive."*

Neurofibromas and plexiforms were seen as particularly challenging. People talked about covering neurofibromas if possible, partly in order to manage the reactions of others *"Yeah in the winter it used to be all right because obviously it was covered but it used to be in the like you know summer months um but I always used to... obviously it used to upset me but then you think why should I change for other people's, you know, ignorance and, you know, rudeness and horribleness."* (Ros, 24)

Several participants mentioned appearance-related bullying at school and in the local community, typically involving name calling and teasing. Some had neurofibromas removed in response to other people's reactions to them *"because of that bullying I did try and get it removed by laser treatment but um it only worked a little bit and it wasn't worth it and obviously because I got older um... because I got older um it didn't really affect me much because obviously when you get older not many people comment."* (Ros, 24)

In considering 'What is NF1?' and 'What does it mean for me?' participants talked of adolescence being a time of change, a time of re-evaluating or learning about NF1 as they came to understand what it was, what it might mean for them and the impact that it might have on their lives in the future. Several commented that when they were younger NF1 seemed of little consequence but it had a greater impact across their life as they got older and through secondary school.

*"In primary school, I remember I still think about it now and I'm just like really surprised that I wasn't ever bullied because of it but I wasn't ever made to feel judged and stuff ....so I never felt like I couldn't talk about it with people and I never felt like people didn't understand it was just something that was there it was never really like a massive deal"* (Sarah, 23).

#### **4.3.3 Theme 2: Relationships and Reactions**

Participants felt their relationships were affected by NF1 in a myriad of ways. This was discussed with reference to (a) *Family*, (b) *Friends* and (c) *Other people's reactions*

##### **Family**

As a genetic condition NF1 can have a significant impact on families in terms of potential inheritance. NF1 was described as a 'family thing' by several participants meaning a diagnosis impacted on the whole family. For some, families were seen to understand the condition, and were an important source of support and information *"Yeah I asked my mum and dad a lot because they experienced it worst than I did..... Normally after a conversation I am ok about it really"* (Robert, 20). Additionally, family members who had NF1 themselves were thought to understand the condition and its impact more than those without it *"...we can compare things, you know and discuss with each other, you know different things but, and then I think myself and my dad can understand each other more, where you know people without NF can't really, you know they think they understand but you know, no I don't want to you know, no offence to them but they don't really"* (Ros, 24).

For others NF1 was simply never discussed within the family, and some participants felt their parents had little understanding of the condition.

*“My Mum doesn’t really, she’s not very well educated and stuff and she didn’t know much about it herself and like for the first, I don’t know how many years of my life I was saying ‘Neuro-fibromatosis’ wrong. I don’t know what I was calling it. Something else, because my Mum had been calling it something else, so obviously, didn’t really know much about it and stuff.” (Sarah, 23)*

In addition to its current impact, NF1 potentially impacts on future family life due to the possibility of children inheriting the condition *“.....the thing that was really horrible that I found out, at the time it was anyway, was about the impact of NF1 on my ability to have children” (Sarah, 23)*. Some people had discussed reproductive decision making with health professionals or had considered future plans. Some were certain that they would not have children in the future *“I always said I am not going to have any of my own children, I’ll adopt.....I don’t want to put anybody, like knowingly, put anybody through the possibility of having to go through everything like all the hospital trips and because knowing that I have had it quite mild but if I had a child that child could have it worse and could possibly like have almost everything associated with NF” (Lucy, 16)*. Others had considered the issue but felt it was something to be considered with a future partner further down the line *“that’s a decision for two” (Daniel, 24)*.

### **Friends**

Consistent with previous research with young people with an altered appearance (eg Williamson et al, 2010) friends were an important source of support, understanding and trust. Some participants found telling friends about NF1 straightforward; *“...it just takes two minutes of conversation and then you don’t need to talk about it again.....doesn’t really bother me when people ask; I’m quite open with it. I’m happy to talk about it. I don’t feel uncomfortable talking about it’ (Mark, 21)*

However, others were adamant that NF1 was a matter that they didn’t want to discuss with others; whilst Ros discussed others’ reactions to her appearance she felt quite strongly that the diagnosis of NF1 was private and as such did not share details of her condition *“none of my friends know I have got NF.....Nobody knows, not even like my best friend, nobody knows, it’s personal to myself’ (Ros, 24)*.

A feeling of trust was an important factor in whether or not people shared their NF1 diagnosis with others, *‘I’ll tell if I knew them like I could trust them” (Tina, 21)* *“if I get to know them a lot better or get to trust them I will probably tell them but not straight off” (Robert, 20)*.

Age at diagnosis also seemed to impact on how comfortable people felt about sharing their diagnosis with others. Those who had received their diagnosis later in life reported that telling others was more challenging, whilst participants who had been diagnosed at a very young age reflected that many of their friends had always known about it and there was never a specific moment when NF1 had to be explained, it was simply part of who they were.

### Other people's reactions

Participants discussed their experiences of managing situations where strangers had stared at them or asked questions about their appearance. Some found this distressing, *"I am always scared that people are looking at you, are they staring at you?"* (Tina, 21) *'I was on holiday and I seen them looking at me and they were whispering and looking at me'* (Joanna, 17) whilst others had learnt to manage it *"I've never really been too uncomfortable because I've grown up with it, so from an early age I've always been used to having to say something."* (Mark, 21).

These day-to-day interactions with other people were typically discussed in terms of the noticeability of NF1. Some young people felt noticeability might be helpful as others would see this as a sign that support may be needed, *'...it's not taken as seriously as if I had loads of growths which were obvious'* (Lucy, 16). However noticeability was also described as challenging particularly when others asked unsolicited questions or stared at them. Participants pointed out that if they responded with 'it's a neurofibroma' or 'I have NF1' others are unlikely to know what that meant and long explanations were needed. Several young people wanted an easier answer to such questions or for there to be greater awareness of NF1 in general so that the questions might be asked less often. Others did not want to answer and would make up explanations or excuses for their neurofibromas and scars *"I just tell people I got stabbed"* (Tina, 21).

Several participants reflected that their concerns about other people's reactions to them could make social situations difficult, and in some circumstances cause them to avoid interacting with others; *"...when I meet new people, there is immediately this thing where, oh she walks a bit different, her legs are a bit weird, she's just tripped over her own feet, like you know, and it's like I just feel immediately awkward about all of those things and I think.....just screw it I won't even bother trying to talk to anybody."* (Sarah, 23)

Consistent with previous research on severity of visible difference some of the participants who appeared least visibly affected seemed the most concerned about other people's reactions, whilst some of those with more noticeable NF1 were seemingly very socially confident.

#### 4.3.4 Theme 3: Understanding and misunderstanding

This theme relates to participants' experiences with organisations including healthcare, school, support groups, and the media, and the impact these organisations had on relationships and psychological adjustment.

## Healthcare

Experience of medical systems differed greatly, however an issue that many found particularly challenging was a perceived lack of knowledge and understanding of NF1 within the medical community.

As Ros (24) explains *“you do find quite a lot that you’re the person you know telling doctors about NF and trying to educate them, quite a lot of doctors don’t know about NF.”* She goes on to explain how this makes her feel *“Obviously you are fighting all the time, you know, educating doctors and the majority of the time you think, oh, and when you go to see new doctors you think, oh, they are going to know about NF. Then they don’t even know that you got NF and you have to say what it is and then they go ‘oh right’ but they will see the basics of it, your cafe au lait marks, your plexiforms, your neurofibromas, but they don’t know, you know, how much in depth it is.”*

Others described occasions when health professionals had not heard of NF1 at all. When Tina (21) drew her timeline she added several occasions when health professionals had not understood her condition. She highlighted a specific occasion when she had asked her GP for information and was frustrated that he had to look up the condition, *‘they looked back on my file and said ‘hang on we have got neurofibromatosis here’.....the doctor was reading like a massive big book in front of me and I was like ‘what are you doing mate, come on’*

Some participants felt this perceived lack of knowledge meant they had to become an expert in NF1 and be responsible for their health *“when I talk about things with healthcare professionals, because they see me as the expert I think I come away from there without any real advice”* (Sarah, 23).

Those who described positive relationships with health professionals commented on their perceived expertise and professionalism. In particular, those who had seen specialist nurses, doctors or The Neuro Foundation specialist advisors described being very happy with the advice and care they had received. Experts had a crucial role to play in putting information into context *“....so I got in touch with her {specialist advisor} and she dealt with it all and told the GP what to do and sent him NF guidelines to follow”* (Ros, 24). It was very important to participants that knowledgeable health professionals were available to give accurate and reliable information and advice on NF1.

## Education

Some participants had positive experiences of school and further and higher education. However, others talked of a ‘battle’ to get the support they needed and to have their NF1 and any associated learning difficulties recognised, and felt this had impacted negatively on their education. Some felt they had achieved well despite little support. In discussing this theme it is important to note that participants had very different educational experiences. Some had been to schools for children with additional learning needs whilst some had attended mainstream schools, some went onto lifeskills

courses at college and others had completed undergraduate degrees. Two participants had post graduate qualifications and two more were planning postgraduate study. There did not appear to be an obvious relationship between those who did or did not have learning difficulties and their experiences at school. Some people with specific difficulties disliked school and some were happy, whilst people without specific learning difficulties also fell into both categories.

Generally, participants felt that having support that met their individual and changing needs was critical. Many were very angry about their experiences at school and felt it was often due to a lack of awareness of NF1, although having a NF1 specialist contact the school often completely changed the situation; *'I struggled all the way through primary school up until secondary school it was only until secondary school where my paediatrician and he stepped in and said this girl needs, you know, help in her work and he arranged for an educational psychologist um and that's when I got found out that I obviously had dyslexia'* (Ros, 24). Tina (21) felt that not knowing she had neurofibromatosis had impacted negatively on her education *"...so I never knew what was going on in my head at school I didn't know why I was so bad at school and kept getting into trouble."*

Positive experience of school seemed to be related to the recognition of any specific learning difficulties and availability of targeted support. Many of those who received good support at school felt this was due to their parents being very proactive when they were young which meant, as Mark (21) explains, *"people were generally quite understanding.....so it was always easy"* Robert (20) also enjoyed school *"I went to two really good schools and they both helped me really well."* Daniel (24) went to a mainstream school and received in-class support. He explains *"I think I did ok with school I got good grades, I went to University, I have a job now so I think it didn't affect me that much"*.

Physical Education (PE) in school was discussed by many participants with the general opinion being summed up by Robert (20) as *"never really enjoyed PE much"*. Most described feeling uncomfortable changing in front of others and worried about what people would think and say. Lucy (16) explains *"I think the only real time where it sort of um... in school where I became uncomfortable was in the changing rooms, changing for PE... we couldn't really get in early because um the changing rooms were always locked but I tried to change quickly."*

### **Support Groups**

Participants had very different experiences with support services and sought support from different avenues. For some, as discussed earlier, support was very much provided by the family or a particular friend. However others talked of religious support and the support from the local community. Some were involved in social activities and groups for specific symptoms, such as ASD or ADHD. Some had looked into online support and a few had sought professional psychological support.

The Neuro Foundation was a source of support for some of the young people in this study. A few had been to camps run by the organisation, which they described as being supportive, particularly because they were designed for their age group, giving them an opportunity to meet others of a similar age and experience to themselves. Those that had been to meetings or contacted advisors were positive about the support *“The NF association is fantastic, you know, they have got us through, you know, through the years they’ve you know they’re our big support link”* (Ros, 24).

Some felt that support groups were more for parents or for those most severely affected *“I wouldn’t want to go to a support group because I wouldn’t want to inter-act with people who had had it much much worse off than me and then I’d kind of feel guilty for being there or I’d just feel like oh my God that’s going to happen to me”* (Sarah, 23).

Some participants preferred to use online support rather than face-to-face groups, and as shown in the following quote, were sometimes pleased to speak to others without the need to focus on their condition. *“I used the instant messaging thing which was really good because there would be loads of people there and you’d be talking to lots of different people and I’d kind of just go on and wouldn’t always really talk about NF it would just be a shared understanding”* (Sarah, 23).

Participants who had met up with others with NF1, either in person or virtually, underlined that there is a shared understanding that comes from having NF1, *“Well, it was a while back when I met my friend and obviously she was there with her mum and I was just chatting to her and then she was having difficulty, you know, with work and she said to me ‘oh do you experience this’ and I said ‘oh yes’ and she says ‘well what do you do?’ and I said ‘well I do this, this and this’ and she said ‘oh right’ and her mum, you know, got quite upset because obviously she didn’t see it as a problem. But I knew straight away what she was, you know, meaning.”* (Ros, 24).

Some were unsure of how to access support. *“I was trying to find support services but I can’t find nothing”* (Tina, 21) and others were interested in meeting other people their own age with NF1 or other chronic health conditions but were unsure how to do this. Participants wanted practical support and advice on managing NF1 that was age specific but were unsure of where this support would come from or how it could be delivered.

## **Media**

The representation of NF1 in the media (such as the internet, television and in print) was important to many participants. Most felt it is misunderstood or unknown to the general population, and worried about the quality of information available, and the negative and pathologising way it is portrayed. *“if I see any articles regarding NF, 9 times out of 10 they got it wrong. That really winds me up.”* (Daniel, 24).

For many the internet is a source of information regarding NF1 but one that is often unreliable, *"I don't trust the internet. If you see it on the internet they always show like pictures of real like severe conditions I guess like and if people do want to know about it, I'd rather they look through the association rather than just Google it because you don't um understand enough of it"* (Mark, 21). Many others agreed that care was needed when looking for information on NF. *"I don't look on the internet because um, because I can remember doing it when I was younger and I scared myself and the paediatrician said never, ever look on the internet, look on The Neuro Foundation and that's all I look at is The Neuro Foundation."* (Ros, 24).

In addition to finding out about NF1 for themselves, participants were also aware that friends and family may use the internet or other media such as television programmes and thereby also find information that could be inaccurate. Talking about the possible impact at work, Daniel (24) explained that he avoids telling potential employers about his NF1 as *"I wouldn't want to say NF because my fear is if they go onto Google or Wikipedia and then find this kid has varying disabilities this kid may have a fit and I don't want them to think that."*

Participants mentioned that programmes on television about NF1 tended to show very extreme examples of the condition and often failed to explain the diversity of the condition. This could impact on how friends and family understand it *"that...is really horrible when you're growing up because you're trying to pass yourself off as a normal person when you're, when younger, and then to think oh my gosh that person has that and you know, worrying what if you're.... what if my friends have read that and thought oh that's what (Sarah) has got as well, that Elephant Man disease or whatever. That was, that was really hard because I'm trying to.... 'cos in.... 'cos I was trying to cope with the things that made me different and trying to make myself as normal as possible and deal with all these things that made me different and then to have those horrible stories like thrown in the mix."* (Sarah, 23)

#### 4.4 Discussion

The findings of the current study support previous suggestions (Ferner et al, 2007; Ablon, 1999) that NF1 may be particularly challenging due to its unpredictable nature and its impact on appearance and social interactions. Young people face these challenges within a society that holds a robust belief about the importance of appearance and the stereotypical view that "what is beautiful is good" (Dion et al, 1972) and during a life stage when body image concerns are likely to be emphasized (Smolak, 2004). Additionally, the findings suggest the lack (or perceived lack) of awareness and understanding of NF1 may make adjustment all the more complicated.

Whilst participants described the challenges they faced as a result of having NF1 (in particular other people's reactions to visible signs on their skin, their concern over the uncertainty of changes and the general lack of awareness of the condition), it is important not to pathologise the experiences of people living with an unusual or altered appearance (Egan et al, 2011; Rumsey et al, 2004) and to

highlight that the participants in this study displayed considerable resilience and were living positively with NF1 despite facing numerous challenges.

As suggested in previous research (Ferner et al, 2007; Ablon, 1999) adolescence was, for many, a time during which the condition became a more prominent part of their life, partly due to physical changes (condition specific or pubertal), but also as they reconceptualised their understanding of the condition. This reconceptualisation, alongside the reported desire for information as symptoms change, suggests adolescence may be a time during which availability of good quality information and access to specialists is crucial.

Young people in this study called for a greater awareness of NF1 amongst health and education professionals and the general public. As has been suggested previously, participants felt this would be helpful (Dheensa & Williams, 2009) and that distressing mistruths such as the persistent (inaccurate) link to 'the elephant man' should be challenged (Ablon, 1999). In particular young people's knowledge and perceptions of NF1 were often linked to experiences with health professionals.

In line with the findings of studies with young people with other appearance-altering conditions, participants valued social support from friends, family and support groups, as well as practical information (Williamson et al, 2010; Thompson & Broom, 2009; Thompson & Kent, 2001) to help them manage other people's reactions to their appearance. Social skills training has been shown to be beneficial for many people with a visible difference (Rumsey & Harcourt, 2007; Clarke, 1999) and adolescence has been found to be a transitional point with regards to young people's social skills in studies with other conditions (Pitt, 2009). Given that young people with NF1 may have poorer social skills than many of their peers (Barton & North, 2007); social skills training would seem to be a potentially supportive and beneficial intervention.

A critical evaluation of methods used within this thesis is provided in the final discussion chapter. However at this point it is useful to highlight that while visual methods, in the form of timelines were planned for use in this study, the interest from participants in using these methods varied. The information that participants were sent prior to interviews included details of using timelines within interviews, (see appendix 6). Whilst some participants were keen to use this method others preferred to talk about experience without using a timeline. A few young people mentioned when arranging interviews that they were 'not very arty' and this seemed to be a concern. Reassurances were given that timelines could be used, but did not have to be. In total three people used timelines, feedback from these individuals was that they were useful and interesting. There were specific reasons as to why time lines were not appropriate across all interviews. One interview was by phone which meant that visual methods were not applicable while another was in a very public space (as chosen by the participant) and drawing on an A3 piece of paper would have been uncomfortable due to space and also other people walking past (although in hindsight the timeline could have been adapted). The

information about the use of timelines also mentioned photographs; interestingly three people bought photos and albums of pictures to discuss but seemed less keen on using timelines. In these circumstances photos were discussed within interviews, they were generally used by participants to illustrate points, for instance in describing their appearance at certain points or when discussing an operation.

In conclusion, it seems that whilst young people describe varied experiences of NF1, central to their accounts of living with the condition and their general wellbeing are 3 key areas:

- First, general feelings about their appearance; across interviews young people discussed their actual appearance as well as managing the uncertainty of possible future changes. Appearance was discussed in terms of NF1 as well as the general importance of appearance both to self and society as a whole. It is important to note that how visible or noticeable NF1 was did not appear to be the most important aspect of discussions of appearance.
- Second, management of social situations and other's responses to visible differences were clearly an important aspect of experiencing life with NF1 for the young people interviewed. This was discussed by all participants in many different ways.
- Third, understanding NF1, or perceptions of the condition, were discussed in terms of what it meant to an individual (and how this may change) as well as understanding of NF1 by others, including school, health professionals and general public/media's.

These three areas, (see figure 5) were used as the basis for developing the questionnaire used in the survey in stage 2 of this thesis. This development and the choice of questions and measures to investigate each identified area are discussed in Chapter 7.

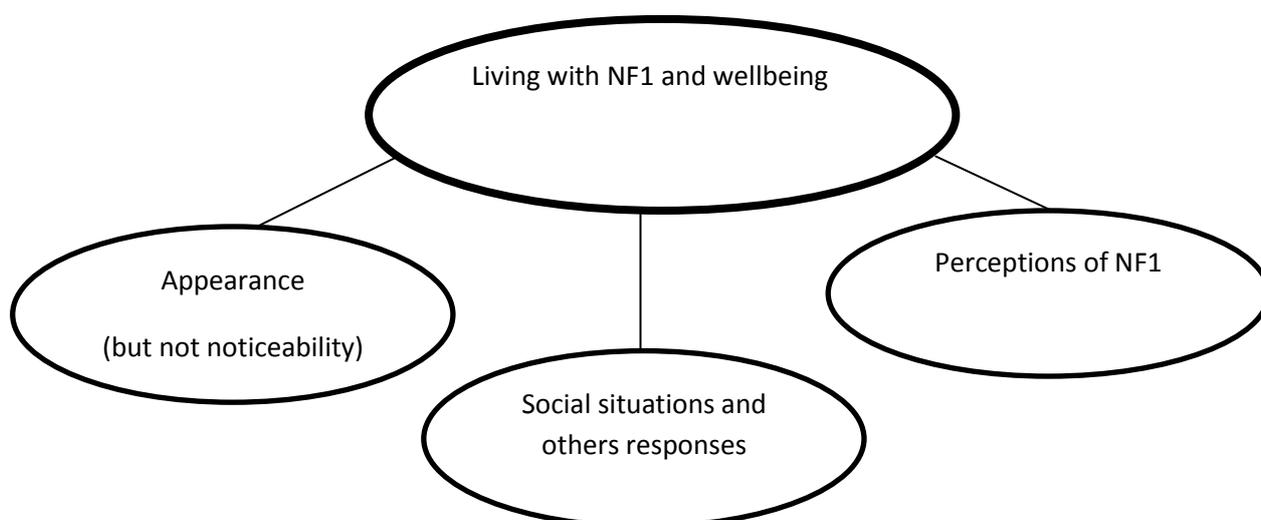


Figure 5: Key aspects of young people's experience of NF1

## **Chapter 5: Interviews with parents**

This chapter details exploratory interviews undertaken with parents of young people with NF1. It describes the study design, interview process, and data analysis.

### **5.1 Aims**

There were two aims of this study:

1. To obtain a comprehensive, in-depth understanding of parents' perceptions of the role that appearance plays within their child's experience of living with NF1
2. To use this understanding to identify areas to be investigated in a subsequent survey

### **5.2 Method**

#### **5.2.1 Design**

The rationale for taking an exploratory qualitative approach and for including parents in this research is outlined in Chapter 3

#### **5.2.2 Ethical Approval**

The study was approved by the Research Ethics Committee of the Faculty of Health and Life Sciences at The University of the West of England, Bristol.

#### **5.2.3 Recruitment**

Recruitment took place alongside recruitment for the young people's interview study. It was made clear in the study information sheets (see appendix 4) and in conversations with potential participants that parents and young people in the same family did not both have to take part, but could if they wished to. Recruitment took place as previously discussed in Chapter 4. As in the previous study participants were recruited until saturation point was deemed to have been reached.

The inclusion criterion was parents who had a child diagnosed with NF1 aged between 14-24 (inclusive) who had an understanding of English sufficient to take part in an interview conducted in English, living in the UK. Parents did not have to have a diagnosis of NF1 themselves although having a diagnosis did not exclude them from participation.

#### **5.2.4 Participants**

Seven parents responded to adverts for the study and all were interviewed: four reported their child's NF1 was new to the family, one had NF1 themselves and two reported that their partner/former

partner had the condition. Two participants (Rachel and Simon) were married to one another and were joint parents of a child with NF1. Four interviewees were recruited from an advert in a Neuro Foundation newsletter, the remainder through online forums and The Neuro Foundation facebook page. All the parents interviewed were parents of young people interviewed in the previous chapter. See table 2 for participant details; pseudonyms are used throughout.

<b>Participant Pseudonym</b>	<b>Gender of child with NF1</b>	<b>Age of child with NF1</b>	<b>Do others in immediate family have NF1? (aside from child)</b>
Gwen	Female	17	No
Michael	Male	20	No
Ellen	Female	24	Yes (child's father)
Angela	Female	14	Yes (Self, child's mother)
Linda	Female	16	Yes (child's father)
Rachel	Male	21	No
Simon	Male	21	No

Table 2: Interviews with parents: Participant demographic details

### 5.2.5 Interview guide

An interview guide was developed through the literature reviewed in Chapter 2. Topics included diagnosis and management of NF1, and the impact of the condition on both the family and the child with the condition. The guide can be found in appendix 8. The exploratory nature of the study and the desire to put people at ease meant that participants were encouraged to take a lead in the interview and to tell their story in their words, and in their own way and time.

### 5.2.6 Procedure and data analysis

The procedure for arranging the interview and the manner in which the interview was conducted was as detailed in the previous chapter (section 4.2.6). Ellen was interviewed by phone, the remainder of interviews were conducted in the participants' homes. Interviews lasted between 30 minutes and one hour. Geographically, participants lived in Northern Ireland, Wales, Northern England, the South West, the South East and Southern England. The face-to-face interviews took place in participants' homes (n= 6), the person who was interviewed by telephone was in their home. Interviews were carried out alongside the young people's interview study and analysis took place after the analysis of young people's interviews. The method of data analysis, thematic analysis, was as detailed previously (section 4.2.7, an example of coding can be found in appendix 16).

## 5.3 Findings

### 5.3.1 Themes

Initial reading of the transcripts revealed that parents had mixed experiences. However it became apparent that whilst the individual detailed experiences were very different there were important similarities in the types of experiences they described, their concerns for their children and the support they wanted. The data was ultimately organised into three key themes, with subthemes as shown in figure 6.

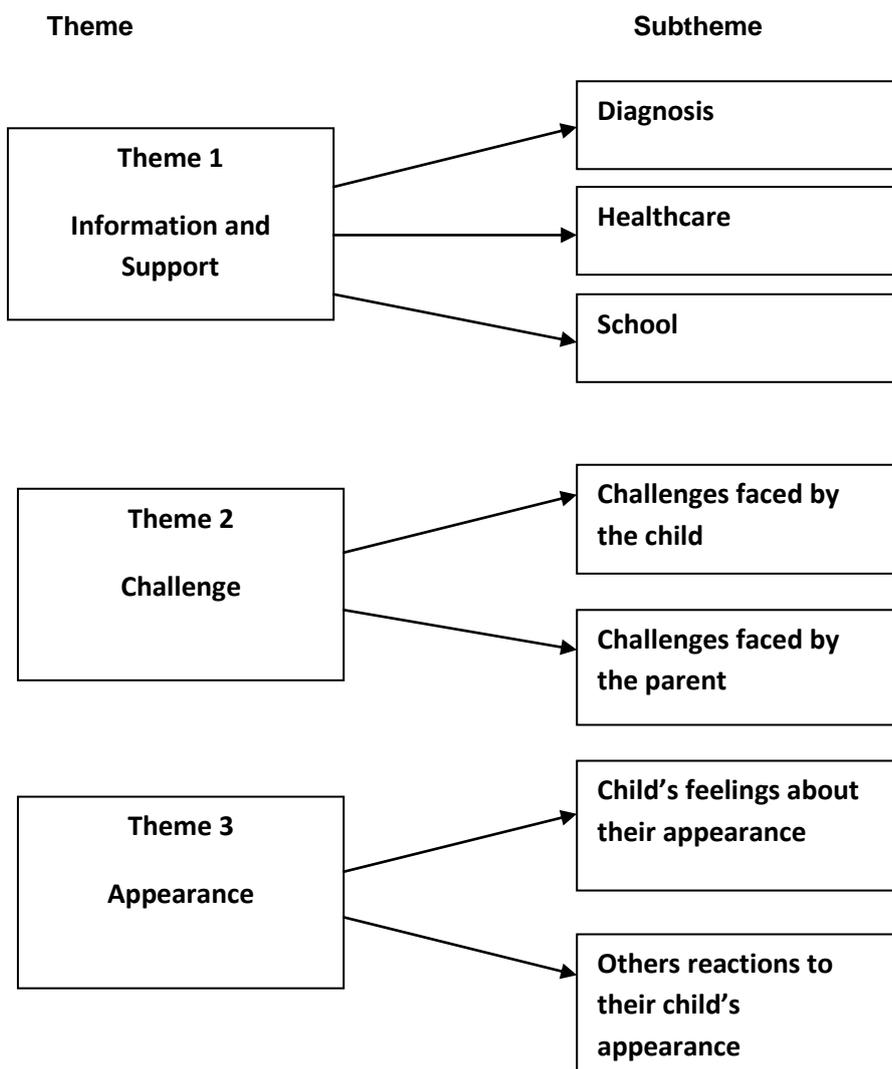


Figure 6: Thematic map of findings from interviews with parents

### 5.3.2 Theme 1: Information and support

Throughout the interviews, participants talked about the importance of information and support for themselves and their child. This was discussed in terms of when children were first diagnosed, and specifically in relation to interactions with school and health professionals.

#### Diagnosis

Participants explained that they generally knew nothing or very little about NF1 when their child was first diagnosed and described the diagnosis as a shock, which caused great uncertainty;

*“It was a shock it took us 3 or 4 years to get used to it really” (Ellen)*

*“I think your big concern is just what does the future hold. So, how bad can it be? How mild can it be? Where is {my child} going to fit amidst those two extremes?” (Simon)*

*“It was completely new, yeah obviously it’s one of those....it’s not a sexy disease is it? .....There is no typical scenario with NF so we just didn’t know where it was going to go” (Michael)*

Participants talked of diagnosis being a time of information seeking and many looked to the Neurofibromatosis Association (now The Neuro Foundation), medical professionals or the internet. Experiences were both positive and negative, although most people felt the first point of contact should be The Neuro Foundation.

*“they’ve {The Neuro Foundation} been really good support right from the start” (Rachel)*

Some participants highlighted the need to take time to learn about NF1 as too much information at once was overwhelming.

*“I didn’t really worry at that stage {diagnosis} because I could not see any lumps or tumors it was just these birthmarks and I thought which was fine but then I went on the internet and read up.....which is the wrong thing to do .....then I got quite upset is this what her life’s going to be?” (Gwen)*

*“I think you read the leaflet and you kind of think ooh there’s a lot of information in there and some things you know are negative.....it wasn’t really until {my daughter} went to preschool that yeah at that point then I think we called on the Neurofibromatosis association and a very nice guy came..the support worker and we saw him at home” (Angela)*

When diagnosis was made later in a child’s life it was sometimes a relief. As one parent explained, her daughter was having difficulty at school and receiving a diagnosis and information about NF1 meant she felt empowered in asking the school to support her child.

*“it’s good because it means that you can now go and say you will have to give us this extra help, you have to do this because we have proof that if a child has got NF, we have got all the literature from the NF association that says, a child with NF might have so and so difficulty with whatever, so you have to do something. Whereas before that they were just brushing it off” (Ellen)*

## **Healthcare**

Participants described very mixed experiences with health professionals. Some described challenging situations where professionals had not heard of NF1, or misunderstood it.

For example Gwen described taking her daughter to a dentist who questioned, in front of a group of people, what she had been feeding her daughter and told her it was disgraceful that her daughter needed to have seven teeth removed. She explained a subsequent visit to a NF specialist showed a tumor growing on her daughter’s gum which had caused the damage.

*“.....there was a tumour in her gum that was attacking it and that’s why she had to get them seven teeth and I knew myself that I was right and I could argue back with her that I was not feeding her junk food and sweets and coke because her diet was restricted it was not everything within her mouth and that made me angry... and it made me think I wasn’t a good mother we are all going to know that this child needed seven teeth removed and it’s not. I mean she humiliated me in front of those people.” (Gwen)*

Other parents also discussed being unsure of how to manage difficult situations where health professionals had misunderstood the condition. As Ellen explained health professionals’ attitudes and reactions were highly significant, and were still remembered years later.

*“.....actually a nurse, I can’t remember who it was it was a school nurse maybe....yes I think it was when she was having her meningitis injection or having some bloods taken she said ‘oh what’s that?’ And she was a nurse which, even if a nurse doesn’t know you would hope that they would say something in a better way than that” (Ellen)*

Experiences such as these seemed to make parents feel frustrated and angry and, for some, led to an assumption that health professionals would not understand. Some discussed feeling the only people they could listen to and trust were specialists.

*“My GP can’t tell me anything and that’s why I don’t bother I just can’t... there’s no point” (Gwen)*

Those who had contact with NF1 specialists had very positive experiences, describing their consultants and nurses in glowing terms, there was a strong sense of being able to trust and rely on information given by specialists, and parents described feeling reassured that they were receiving

good quality care and advice. Parents wanted to feel their child had the best possible care and seeing doctors looking unsure, making mistakes or looking up symptoms was unnerving and worrying.

*“You look at the information and sometimes it focuses on the extreme so you get a little scared by it and just think actually until I know, I won’t take any information off the internet. I’ll wait to hear what the specialists that see {my child} have to say about it. ....the doctor that we saw seemed to have a good understanding of the condition rather than fumbling through books and not really knowing.”*  
(Simon)

### **School**

The majority of parents commented that obtaining educational support that met their child’s needs was their greatest challenge. Ellen’s response *“fighting the schools”* sums up many parents’ responses. Parents often described trying to get support as a ‘struggle’ and a ‘battle’. Some felt that their child’s school had let them down; others described how it had been difficult to obtain support.

*“It was horrendous battle because when I first, it’s a local primary school and when I first contacted them they were saying oh he’ll be fine. We’ve got special needs here and I said no, you know this is what he’s got. Oh no he’ll be fine, it will be fine, he won’t need any help and I thought no, he will need help because I knew because I had a friend who was a teacher and he used to go up there, we used, before he started school when he was at playschool, it was quite a struggle at playschool. And I thought no, so um it was really hard, it was a big battle”* (Rachel)

There was a strong feeling that schools did not understand NF1 and the learning difficulties that could be associated with it. Parents reflected that the variability of the condition made it difficult to explain and that very few schools or teachers knew about it. Angela explained that her daughter had an ASD diagnosis and she felt that this, because it was better known and understood by schools, was the reason she received support, not the NF1:

*“There is probably more support for the autism than there is for you know if I just said oh they just have neurofibromatosis and they have you know perhaps some difficulties with social skills and things. You don’t get any support for that but by saying they have autism, actually you know we get the autism outreach are coming to the school and you know and explain to the school how they can be supported. But in a way I feel a bit cheated sometimes but yeah having just a genetic condition almost doesn’t seem to be enough sometimes.”* (Angela)

However this was not the case for all parents and some were happy and felt their child’s school had been supportive and proactive. This was related to parents feeling their child’s needs were being met and in some cases receiving specialist provision:

*“...they were so supportive we didn’t have any problems getting him statemented”* (Michael)

*"I do think the special needs was the best place for {my daughter} in terms of schooling because she was among people who all had difficulties and all had disabilities and it didn't make her any different. So I did think it was good and now she's moved on to Tech and it's the same thing it's a special needs." (Gwen)*

Once support was in place parents described feeling they had to be continually proactive and closely monitor their child's support and attainment to ensure consistency. Simon explained that he or his wife often had to *"...be right on the case, all the time it was just making sure {our son} got what he was entitled to and they {school} had the understanding that they needed about his condition"*

Overall, whether their child received good educational support or not, parents felt a very strong sense of responsibility for their child's education and described monitoring and worrying. It was a clear concern for many; particularly due to the perceived lack of knowledge and recognition of NF1 by schools and teachers.

### **5.3.3 Theme 2: Challenge**

Participants recognised that having NF1 affected their child in many different ways and that there was also an impact on them as a parent, as an individual and on the rest of the family.

#### **Challenges faced by the child**

A few parents worried that their child only had them to talk to about their condition and were concerned that if their child needed more support it may not be available. They described feeling that young people could benefit from psychological input from specialists, but finding this professional support from counsellors or psychologists was challenging. Gwen described a long and difficult path to finding a psychologist for her daughter to talk to about managing surgery and social concerns including bullying:

*"She {daughter} needed support in terms of bullying and in terms of surgery and we got her transferred to a psychologist but it was all my own doing. I mean if I was a parent that didn't know how to deal with this there would be nothing for {my daughter} they weren't coming to us. I have had to seek them..." (Gwen)*

Others explained that, as a parent, they felt that they were left with the responsibility for their child's psychosocial needs. A few were concerned that, as their child grew up, they might not want to talk to a parent and worried about who their child could talk to about their concerns. When asked what could improve their child's experience Linda's response was typical of many;

*"...a counsellor within the health system, definitely ...."(Linda)*

When asked what they felt had helped their child, participants suggested that staying calm, being matter of fact about NF1 whilst also making sure their child had support and building their confidence

were all important. Simon explained that for his son, learning to play an instrument and playing in concerts had been helpful in supporting the development of his self-confidence. Being part of a group and having something to be proud of made him feel positive in himself and gave him an identity as a 'good musician'.

*"I think that's a really important point for any child is that they can be good at something like that, it just gives them that confidence" (Simon)*

Parents also discussed the general concerns they had for their child; this included the way in which their child learnt about NF1, and their concerns for the future, including future romantic relationships, and whether their child would or could have children of their own. A primary concern was balancing a desire for children to grow up to manage their condition and be independent whilst finding it hard to let go after many years of managing a medical condition. Participants felt their child with NF1 possibly needed support more than other children;

*"you want to protect them you don't want them to be labelled with anything or...every mother does that they don't want their children to be hurt but I think that's...I don't know if it is but I feel like it's probably a bit stronger" (Ellen)*

*"My daughter (who does not have NF1) I thought well she'll be fine she's alright yeah but for {son, who has NF1} its harder to let go really.....I think I'll be the one that always says to him, like I did this year, right it's your annual check up, you'd better book in. But I left it to him to go and book it but I think I have to remind him still I don't think that will ever go" (Rachel)*

### **Challenges faced by the parent**

Participants discussed managing and coping with NF1 as an ongoing process. They described going through an early process of information seeking in order to understand the condition, and felt that it was important to be knowledgeable about the condition but also to continue with family life as normally as possible and to be philosophical about the future.

*"you realise that you have to learn with it, face it as it comes..... it {the worst case scenario} may not happen but at least I know what to do for the signs if it can happen you know what to look for. So I am at that stage where I know quite a bit." (Gwen)*

Parents described monitoring their child for symptoms and worrying about the worst case scenario, particularly in the early stages, just after their child was diagnosed, or when they had not seen their child for a while, for example while they were at university,

*"every time you see {child} you just sort of, well especially if he's been away, it's like well how are your lumps and your bumps and what, is there anything that's hurting and then maybe you have a look at it and you think oh yeah ok" (Simon)*

As time went by parents described relaxing day-to-day, but explained how any new symptom or a specific event could take them back to how they had felt at diagnosis.

*“You just get on with your life but then something happens and you have a big upset” (Ellen)*

*““You know the condition is very evident so it doesn’t ever go away but I think the annual check and maybe just even the newsletter {Neuro Foundation publication} coming just kind of triggers it again” (Simon)*

Ellen explained that, now her child is 24, she sometimes forgets how difficult it was when her child was first diagnosed, but that seeing articles on NF1 or hearing about other people’s difficulties reminds her of previous events and emotions.

*“...it {newspaper article about NF} brings back all the struggles that you had because now that I mean {my child} is 24 those problems have faded a bit. You forget about them, you cope with them at the time and you forget about them but that brings it back to you....you forget how you coped with it that it was even there even. You block it out; yeah yeah definitely.....I don’t know how I did it but you just do it don’t you?” (Ellen)*

There seemed to be a general agreement by parents that it is important to get on with making life as normal as possible for their child. Several parents mentioned a dislike of the word ‘coping’, partly because they felt it singled them out as different, having to cope means there is a problem and many highlighted that they wanted their child to be seen as any other child would be. Additionally, some parents felt uncomfortable with being told they were coping as they felt they had no choice but to manage the situation they were in. As Michael explains, ‘not coping’ does not seem like an alternative therefore ‘coping’ is just getting on with it.

*““you know when you are out people say how can you cope? How do you think you do? You can’t just say I can’t cope and walk away” (Michael)*

Participants also talked of comparing their situation to others and looking for the positives in their circumstances and making downward social comparisons; ie comparing themselves to others in a more difficult or challenging position. Several used examples where they compared their child to another with what they perceived to be a more difficult or threatening condition;

*“It’s quite interesting, there was a family who lived on the estate but moved on and their child had cystic fibrosis and one day we were up the pub in the garden and chatting away and we sort of went through our experiences and we came away thinking ‘oh we thought we had it bad you know obviously they got ongoing issues and such.’ And then we bumped into them again a few weeks later and apparently they went away and did the same. So you know its...yeah maybe you think you’ve got it hard but other people have got it hard” (Michael)*

Parents differed in terms of how much they wanted to get involved with support groups and how beneficial they felt they were for them and their child. Some felt it could be distressing to attend groups in person as they see people who appear to be greatly affected by NF1 and explained that this can cause concerns and raise issues that had not yet been considered.

*“,,there were a lot of people absolutely covered.....and I think it's like oh I don't think we'll go again I think it was almost too frightening” (Rachel)*

*“It's that balance of getting involved and talking to people in similar situations and then not getting too dragged down by other peoples conditions” (Simon)*

Some parents felt support groups were more useful for those most affected by NF1 and worried that groups concentrated on the worst case scenario too often. Some felt newsletters, websites and information could be too negative and complained of a lack of positive stories, and examples of managing the challenges presented by NF1 well. Several parents mentioned that although NF1 is a variable condition and many have it mildly, this is not reflected in the images support groups use. Simon explained how he felt this could be addressed;

*“I mean what they could do in a way is where you've got somebody who's got it mild.....is actually make more of a role model of people like so you can say to parents whose children have just been diagnosed, look here's a guy or girl who goes to university, they've done this, they've done this, it hasn't actually affected their life” (Simon)*

Some felt that support groups were a good way to meet other parents and talk to others who have faced similar situations thus enabling parents to discuss strategies, experiences and feel less alone. Several parents described the relief of talking to other parents and finding issues, particularly regarding appearance, learning difficulties and behaviour management were common to others. People described how this made them feel their experience was more normal. Ellen explained that a shared understanding can be comforting;

*“It's good to see other people sometimes, sometimes it's nice to sit and talk” (Ellen)*

Some participants explained that NF1 had brought them and their wider family closer and this was described as being very supportive, while others talked of family members not understanding NF1 or what they were going through. The impact of NF1 on family relationships was often complicated.

*“I don't know if it made us closer or if we're close in spite of NF1” (Michael)*

*“I blocked my parents and my family out because they didn't understand and I was hurt by their comments, not that they were cruel, but they would say 'oh it's all right' and when it wasn't all right” (Ellen)*

Participants discussed feeling they were forced into behaving in a certain way and had to become their child's advocate; they felt they had to be in control of medical issues and appointments alongside managing school support and worrying about their child's emotional wellbeing and happiness. At times this meant having to be proactive and having to fight against hospital and schools. Parents explained that they worried that this could give them the label of being a 'fussy' or 'bad' parent and many felt torn between wanting to be a 'good' parent to their child and being a 'good' parent with health professionals and schools.

Gwen explained that her daughter sees different medical specialists as well as her GP and she sometimes finds it difficult to know who to contact and when.

*"I don't want to sound like a fussy mother, there she goes again going to the doctor if anything crops up but I prefer to go to them because sometimes my GP has said ah your fussy and I have always remembered that word fussy mother and I am not, I am just concerned."* (Gwen)

While, in this instance, Gwen explains her concern relates specifically to a GP using the term 'fussy', other parents described very similar feelings and felt conflicted by concerns of over and under reacting.

Some discussed how little awareness of NF1 there is amongst the general population and felt that if NF1 were better understood by the general population as well as health professionals and educationalists, life would be easier. The 'battle' previously described to get school support and the best medical care was explained as being compounded by a perceived lack of understanding. Parents expressed their frustration that NF1 is not generally known, meaning that they felt they had to keep explaining the condition. They felt that wider recognition of the condition would make their and their child's lives simpler and several compared NF1 to other conditions.

*".....you know what I don't understand I mean you have heard of Down Syndrome and NF – no, no if you said to someone outside my child has Down Syndrome, oh dear they automatically know what's wrong with their child, when you say Neurofibromatosis they don't but it's just as common I mean it should be known"* (Gwen)

### **5.3.4 Theme 3: Appearance**

All the parents talked explicitly about the impact NF1 has, or might have, on their child's appearance, both in response to direct questions about appearance and in response to more general questions. This was discussed in terms of how they thought their child felt about their appearance as well as how they, and their child, managed the reactions of others.

#### **Child's appearance**

Some participants felt appearance concerns were central to their child's experience of NF1 while others felt it had not been an issue for their child. Appearance concerns that parents reported related most often to café au lait marks, plexiforms and neurofibromas. Gwen described how a plexiform on

her daughter's cheek meant she avoided looking in mirrors and explained that her daughter often expressed concerns about her appearance.

*"...she doesn't like mirrors and if she gets new clothes, she loves new clothes, but won't go and look at them on her .....she says 'I wish I didn't have this lump' and she would ask me 'why have I got this lump?' and things like that (Gwen)*

Parents described their child's appearance in terms of what was, or was not, noticeable;

*"She has got a few fibromas.....so she wouldn't have worn a bikini or something like that because then people would have seen. She has got a plexiform but it is on her back so nobody would see that and as well hers isn't as prominent as {her sisters} so she could hide it quite well....the only thing with {daughter} is she has got a large plexiforms on her forearm and she sometimes gets upset about that and people do ask.....most people don't ask now she is older but at school and things people used to ask and she used to say it was a birthmark." (Ellen)*

Parents also explained how difficult it can be for them to see their child looking different to others. For many the appearance changes resulting from NF1 was distressing and caused them great concern.

*"I think even four or five years ago I couldn't talk about it without crying, my beautiful little girl..." (Linda)*

That symptoms cannot always be hidden was upsetting for some parents who felt that if the condition could be camouflaged or, as below, if it were not highly visible, then it would be easier to manage. The very visible nature of some aspects of NF1 meant that it was always apparent to others.

*"it's sad I wish she didn't have it and emotionally yes I do wish I could take it away from her, it's not like any other condition where you have a lump on your arm you can remove it, you can hide..... but because it's her face it's more difficult and it causes problems when she goes outside and I just wish it wasn't on her face" (Gwen)*

Participants talked of weighing up the pros and cons of surgery and having to decide whether a procedure was 'just cosmetic'. Operations for cosmetic reasons were seen as being more superficial than those with a medical justification. Several parents discussed how it was sometimes difficult to know if their decision was justified possibly indicating a concern that worries about appearance could be perceived by others as unimportant. It seemed that although parents described visible signs as very difficult for them and their child, surgical intervention was seen as an extreme intervention and parents worried their concerns might be frivolous.

Michael explained how one cosmetic procedure was particularly difficult for him, his wife and his son to decide upon, and highlights the idea of 'needing' an operation. Where there is a clear medical impact 'need' might be obvious, however when the benefit is cosmetic therefore ultimately psychological it appears harder to justify.

*“On the cosmetic side, the reason why we had the squint done was for cosmetic reasons and that was the one thing we had to talk him into. I mean he really didn’t....In fact to some extent he thought it defined him...it was just before he was transitioning to secondary school and we thought well now’s the time to do it before he goes in and he thought well people know me with the squint will they know me when I haven’t got it and we said of course they will know it’s you .....the photos pre and post squint its quite noticeable you know you just get used to it....and my wife agonised more over that one than the brain surgery because she knew we had to have the brain surgery whereas he didn’t need the squint operation” (Michael)*

### **Others’ reactions to child’s appearance**

For many, the visibility of NF1 meant managing other people’s questions and reactions to their child’s appearance, and supporting their child to do the same. Participants talked about how difficult the visibility of NF1 could be and how people often asked questions at inappropriate times or in insulting ways. However, parents differed in how they would like others to respond to their child. Some felt that others should just ask about their child’s appearance, for example, Rachel explained how she gave information to other parents:

*“....then I would say if you want, I had some little leaflets, if they wanted leaflets and I would, I’d just say to people that it’s one of those conditions that a nodule could grow on any nerve ending, they’re inside, they’re outside the body we don’t quite know what’s going to happen you know he has to have his eyes checked annually and this is why he can’t ride a bike, he finds it difficult to do sport blah, blah....” (Rachel)*

Other parents thought people should never comment on their child’s appearance. Gwen felt quite defensive about the way other people sometimes look at her daughter and ask questions:

*“I would be very defensive; what are you doing don’t stare at my child she doesn’t deserve that.....in Majorca, she {stranger} actually came to me and said um ‘you don’t mind me asking what’s wrong with her eye’ and I said ‘I do mind you asking’ because I thought you are not concerned you’re nosey you didn’t want to get to know me first or get to know {daughter} she just walked past and walked back and I thought she was rude and I told her yes I did mind her asking, there’s a way to break it you just don’t come to somebody in the street and ask what’s wrong with you, that’s my feeling I wouldn’t do that with anyone, I would get to know a person first and if they want to share it fair enough if they don’t let them be...” (Gwen)*

Several parents described similar experiences and discussed concerns that their child needed to manage other people staring and making comments. Some commented on how appearance concerns and the noticeability of the condition became more of an issue during secondary school. Rachel explained that although her son’s condition was not particularly visible she felt he was generally more vulnerable than other children. Several parents like Rachel in the quote below,

mentioned swimming, sports or PE changing rooms as being particular concerns for their child. Day-to-day marks could be hidden but some situations were very exposing.

*“I remember him saying oh when he would get changed for PE, people would say, oh what’s all those on you then? Because he’s got quite a few lumps on his chest” (Rachel)*

However Rachel later explained that her son dealt with questions well and was able to explain that the lumps were just part of a condition. In her view it was important to be open and talk to others about NF1 and she had encouraged her son to do the same.

## 5.4 Discussion

The first aim of this study was to obtain a broad, in-depth understanding of the role of appearance within parents’ day to day experiences of having a child with NF1. The findings presented in this chapter demonstrate that parents of young people with NF1 have varied experiences and that appearance affects many different aspects of their and their child’s life.

In more general terms, parents talked of their child seeing a variety of health professionals, some NF1 specialists, some not. Children had a range of different symptoms of NF1 and associated learning difficulties. They attended mainstream and specialist schools, colleges, university and some had post graduate qualifications. Participants talked of families being both closer and more distant, some discussed keeping the diagnosis private while others wanted to educate and share the diagnosis with others. However while parents in this study had varied experiences of having a child with NF1, in order to positively manage its impact and support their child there were common areas that shaped parents’ understanding and perception of the threat of NF1 to themselves and their child.

Participants talked a great deal about how and when their child was diagnosed and the way in which this happened. Early encounters with professionals and information about NF1, including how it might affect a child’s appearance, seemed to have great meaning and a lasting impact for parents, as highlighted in previous studies (including Crossen et al, 1998; Benjamin et al, 1993). The current study further highlights the value many parents place on specialist involvement in their child’s healthcare, particularly against a backdrop in which they worry that many healthcare professionals do not know about or understand NF1. Furthermore, this study also highlighted the ongoing and changing information needs for their child and themselves. Previous studies investigating the information needs of parents with a chronically ill child have also highlighted the individual, shifting and changing information needs of parents. For instance Hummelinck and Pollock (2006) reported parents’ need for information varied over time and some parents in their study reported difficulties taking in information at diagnosis. Yet, while some parents felt overloaded with information, others wanted more. The authors suggest that over time informational needs may be related to a parent’s confidence in managing their child’s condition. Changing information needs may also reflect a child’s

age and a parent's desire to be an intermediary, providing their child with information throughout childhood (Starke & Moller, 2002). Understanding a child's medical situation has been highlighted as a possible coping strategy for parents of children with other chronic conditions such as juvenile idiopathic arthritis (Cavallo et al, 2009). During these interviews, parents discussed a range of coping strategies, including understanding the condition. Many described an initial period of worrying about the worst case scenario, often just after diagnosis, followed by a period of searching for information and understanding and then described a realisation that life could not function at that level of worry. Parents described how, over time, NF1 became a part of life; although they explained that it was always on their mind and events could be triggers that easily take them back to a very emotional state of being. Similar patterns of adjustment and coping are reported in Grootenhuis and Last's (1997) review of coping and adjustment of parents of children with cancer. Fisher (2001) writes that a change of lifestyle clearly takes place for parents who have a child with a chronic condition. What is less clear is how parents manage the changes that chronic illness brings. Adaptation may be related to informational needs being met (Cavallo, 2009), individual personality traits (Vermaes et al, 2008) social support and optimism (Grootenhuis & Last, 1997).

The reported challenges of having a child with NF1 were often related to others not knowing about or understanding the condition. Several participants talked about difficulties ensuring their child had the support they needed at school; they reported that the condition was not well recognised by teachers. Additionally, parents discussed managing situations where health professionals did not know about or fully understand NF1. Another more general concern was that parents perceived NF1 to be poorly recognised and understood by society as a whole. Participants felt portrayals of the condition within the media were negative and pathologizing and information was often incorrect. Lack of knowledge of the condition may make it more challenging for parents to access support and gain the understanding of those around them.

Participants talked about their child's appearance primarily in terms of worrying about the noticeability of NF1 and the reactions of others. Parents discussed monitoring their child's appearance and checking for any changes, and often discussed noticeability in terms of quantifying how visible the NF1 was. They reported concerns that some people did not know how to interact with their child; there were several stories about strangers staring and asking questions. Some worried about peers at school and there were concerns about teasing and bullying. Several parents talked of having purposely chosen activities and groups for their child to join, in order to enhance their child's social skills and build their confidence. The reactions of others and their child's management of social situations were key issues for these parents. Parental support of young people with an altered appearance has been discussed by researchers including Williamson et al (2010) who found that parents of adolescents who appeared to be coping well with an altered appearance were those who supported their child's integration. Williamson et al also note that parents often want advice on supporting their child to feel positive about their appearance and how to manage any concerns or negative reactions from others.

In conclusion it would seem that whilst parents described varied experiences of having a child with NF1, three key areas are central to the way in which they experience and understand NF1;

- First, parents' perceptions of their child's management of social situations and other people's responses to them were highly relevant to how parents perceived their child's experience of NF1. Any difficulties their child experienced related to NF1 were often social, related to friendships and in particular to managing others' reactions. In addition, parents felt that positive social experience was important in positively managing the impact of NF1.
- Second, parents' individual perceptions of NF1 were relevant to their accounts of their own and their child's experience. Parents identified a range of coping strategies that they found supportive in managing their and their child's experiences of NF1 generally and appearance aspects specifically.
- Third, their child's appearance and in particular the noticeability of their child's condition was highlighted by parents as an important aspect within their and their child's experience. Parents seemed to feel that NF1 that was more visible impacted to a greater degree on their and their child's lives.

These three areas, (see figure 7) were used as the basis for developing the questionnaire used in the second stage of this thesis. This development and the choice of questions and measures to investigate each identified area are discussed in Chapter 7.

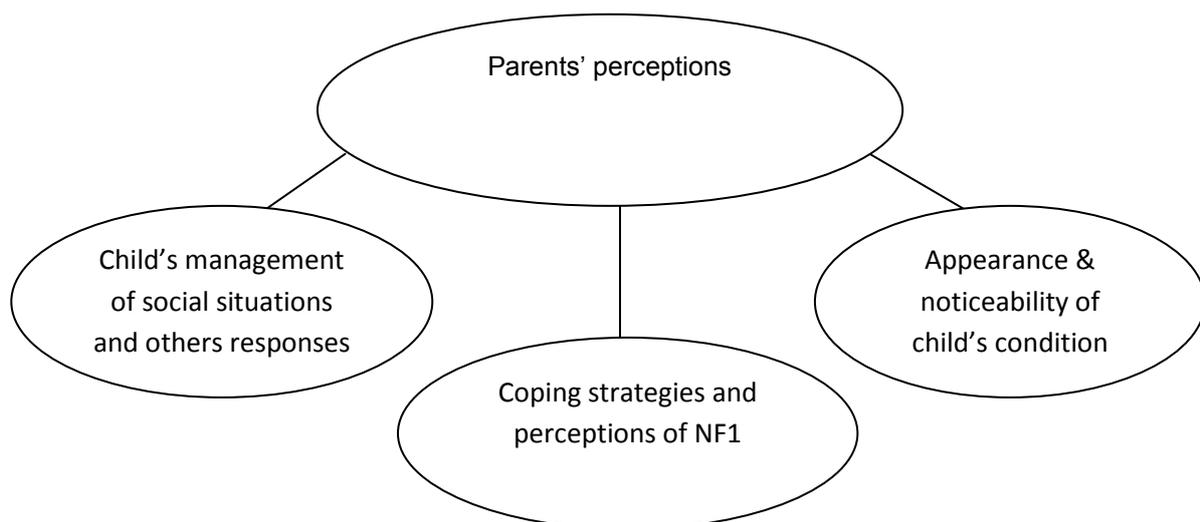


Figure 7: Key aspects of parents' perceptions of their child's experiences of having NF1 and their own experience of parenting a child with NF1

## **Chapter 6: Interviews with health professionals**

This chapter details exploratory interviews undertaken with health professionals who work with young people with NF1 and their families. It describes the study design and interview process and presents analysis of interviews.

### **6.1 Aims**

There were two aims of this study:

1. To obtain a comprehensive, in-depth understanding of health professionals' perceptions of the role that appearance plays within young people's psychosocial experience of living with NF1
2. To use this understanding to identify areas to be investigated in a subsequent survey

### **6.2 Method**

#### **6.2.1 Design**

The inclusion of health professionals reflects the important role some play in young people's experience of NF1. Health professionals who specialise in working within genetic counselling and/or NF1 specifically are uniquely placed to discuss the context of treatment, healthcare structures and management of the condition as well as more individual differences in managing the condition.

#### **6.2.2 Ethical Approval**

The study was approved by the Research Ethics Committee of the Faculty of Health and Life Sciences at The University of the West of England, Bristol. Additionally the NHS National Research Ethics Service (NRES) was contacted and the response advised that NHS ethical approval was not required for the purpose of this study (see appendix 14 for email).

#### **6.2.3 Recruitment and Participants**

Discussions with members of support groups and clinicians who work with people with NF1 identified specialist nurses and genetic counsellors as those best placed to discuss the psychosocial issues young people with NF1 and their families manage on a day-to-day basis. Information about the study was sent to genetic counselling services and to NF1 specialists who were asked to forward details to colleagues who might be interested in taking part.

Initially four female health professionals were interviewed although one participant later withdrew from the study and chose not to give a reason why; as such their data was not included in the analysis.

The three remaining participants were from different areas of the UK, they had nursing/genetic counselling backgrounds and had worked in their field for differing amounts of time; one participant was a genetic counsellor whose work included working with patients with NF1 while the other two participants were highly experienced specialists in NF1. Participants are potentially very easily identifiable by any description of the work they do and as such it was agreed that details of professional roles would not be reported. The pseudonyms of Karen, Ellen and Joanna are used throughout this chapter.

#### **6.2.4 Interview guide**

An interview guide was developed through a review of the literature relating to NF1, appearance, young people and NHS guidelines and practice relating to NF1. The guide can be found in appendix 9; it included questions regarding the health professional's role, the support needs of young people and families, and availability of appropriate support. Participants were asked whether appearance was a concern for young people with NF1, and what were the most common concerns for young people and parents as well as being asked for examples of positive management of the condition. Questions and topics were broad as the aim of the interviews was to identify a range of areas to explore further and to ensure that interviews were participant led.

#### **6.2.5 Procedure**

Potential participants were sent information sheets in advance and were asked to sign a consent form before the start of the interview; they were also informed that they could withdraw their data from the study up until the completion of questionnaires that were based on the interviews (a specific date was given). Interviews all took place in the participants' place of work during the working day at a time convenient to the participant. They began with the interviewer introducing themselves, the project and explaining why the study was taking place. Participants were asked about how they came into the work they were doing and were asked them to explain a little about their role. They were all professionals with experience of discussing their work. Sometimes participants talked about issues that were not part of the interview guide and these were allowed to progress in order to be open to the possibility that these might be previously unexpected areas that needed exploration.

Due to the exploratory nature of the interviews the questions were open and some were interpreted in different ways by different participants. Interviews lasted between 30 and 50 minutes.

#### **6.2.6 Data analysis**

Interviews were transcribed verbatim and detailed field notes were kept by the researcher after each interview. Thematic analysis was chosen to analyse this study, as in the previous two studies.

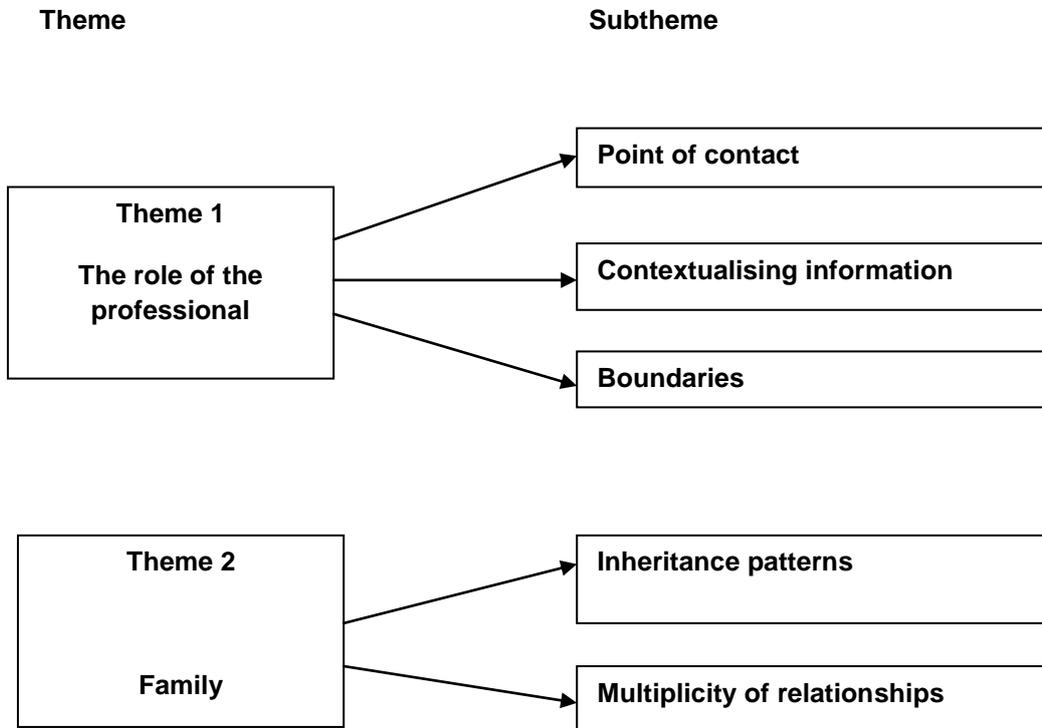
Writing this chapter has entailed some ethical consideration. As previously highlighted, participants are potentially easily recognisable by the comments they have made and the description of the work they do. NF1 research, treatment and support is a close community and participants' observations

may mean others in their field of work are able to identify them. The conflict between presenting detailed full descriptions of participants' accounts and the desire to protect their confidentiality and anonymity in this instance related to what has been labelled 'deductive disclosure' (Kaiser, 2009); referring to instances where the content of quotes could provide clues to the identity of the participant. Kaiser (2009) suggests that in order to address this issue, researchers should discuss the use of the research with participants, including how the research will be disseminated and agree with participants how data will be used in order to ensure consent is informed. The intention with regards to this dilemma, in this instance, was that any comments that identify participants would be anonymised so that the general meaning is kept but the participants' identity/role is not disclosed. The original comment and anonymised comment were shared only within the supervisory team. Lists of quotes were given and the team were asked for feedback to ensure overall meaning had not changed.

### 6.3 Findings

#### 6.3.1 Themes

Analysis revealed four key themes, with subthemes as shown in figure 8 and expanded upon in the sections below.



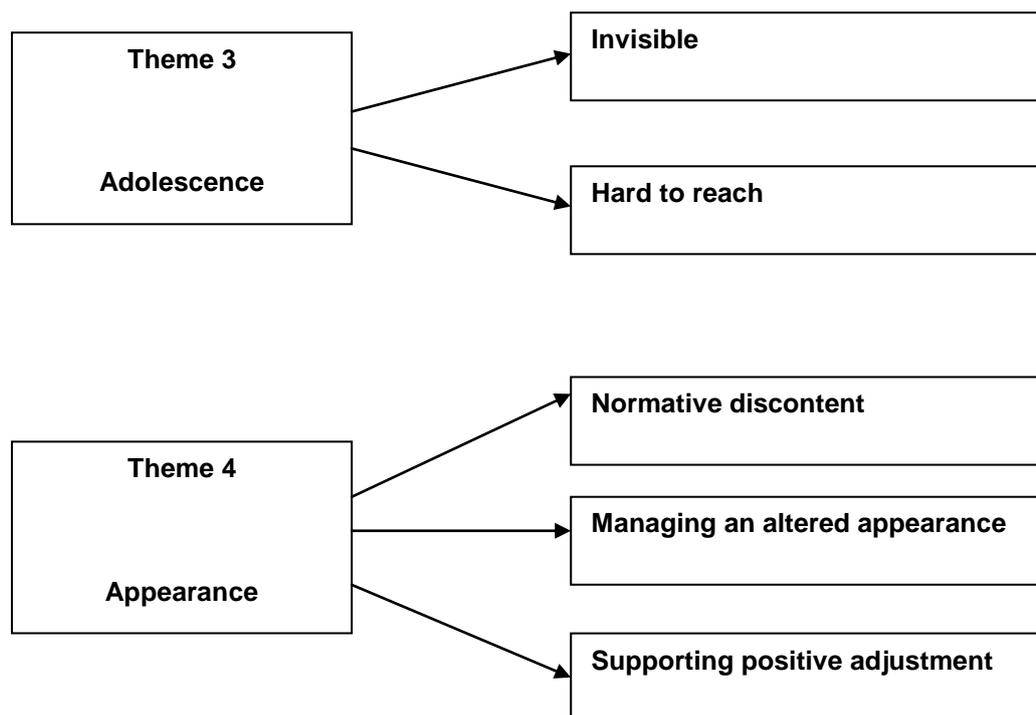


Figure 8: Thematic map of findings from interviews with health professionals

### 6.3.2 Theme 1: The role of the professional

Participants discussed the pragmatics of their work. More philosophically they discussed the aim of their role and the parameters of their position. In discussing their roles there were three subthemes. They described themselves as being a point of contact, they felt an important practical aspect of their role was putting information into context for young people and families and, finally, there were areas that participants felt were outside of their remit or professional experience.

#### Point of contact

Participants identified themselves as often being the initial contact for patients and their families. Whilst they might triage cases and pass them on, an important aspect of their role was being able to explain the condition to new patients. Participants talked of the diverse ways in which people were initially referred to their service and highlighted that diagnosis could be made at different ages and made in many different ways.

Being a point of contact seemed to go beyond initial referrals. Participants described occasions where families had been in touch in order to re-engage with health services after a period of not seeing health professionals for a considerable period of time. They explained that sometimes they were seen as an easier and more approachable avenue than consultants and hospital systems. Participants talked about seeing children, young people and families at different stages, as needed. They may not

see someone for an extended period of time but people would contact them, maybe if their child changed medical teams or moved school, for example.

*“We don’t hold registers....and then re-contact them every so often, we don’t operate in that way. It’s much more that individuals hopefully receive the message that they can come back to us at different points in their lifetime...” (Karen)*

*“...if they’re worried just give us a ring and we’ll see them anytime” (Ellen)*

There was also some discussion of “getting it right” and “finding the balance” and the hope that people would contact them if needed but, as Joanna reflected, this may not work and some people may be reluctant to call.

*“I might think well I’ve given that family my number and I haven’t heard from them and I might hear from other areas that they wish I’d been in touch” (Joanna)*

Another part of being a point of contact was in signposting patients and families towards other support services and supporting patients outside of the medical context. For instance, participants spoke of their role in contacting schools and helping teachers to understand the needs of children and young people with NF1, in this sense they became a contact point for outside agencies.

### **Contextualising information**

Participants talked about making sure that patients, and parents, both got the information they needed and understood that there is some uncertainty within management of NF1 and as such some questions cannot be definitively answered. As Karen summarises: *“how much is it that they don’t know but that is known broadly and how much is things that just, you know, cannot be answered. I think that’s one of the things that I suppose we can try and help to answer - what is there and what can we talk about and what can be known”*

Participants discussed their role in putting information into context. Ellen described a patient commenting on concerns about a recent TV programme and how she tried to reassure them regarding the unusual presentation of the Neurofibromas within the programme *“he said to me ‘did you see that Embarrassing Bodies story?’ and I said yeah and he said ‘what did you think?’ and I said well they were very big neurofibromas”*

In another example Karen explained how specialists can balance information: *“So I think again having that chance to discuss it with a {specialist} who can give it a balanced view of their experience of neurofibromatosis, what the literature says about the condition, hopefully can sort of give them a more balanced view.”*

Participants referred to actively recommending that patients stay away from searching the internet for information regarding NF1 and described the impact that internet searching can have.

*"...they go on the internet and see the extreme; you know cases with multiple neurofibromas and there are maybe plexiforms and they're worried.....I tend to recommend that individuals don't just Google neurofibromatosis." (Karen)*

*"...and then you say don't Google and then the first thing they do when they get home is Google and there's quite a gallery of horrors, as you will know the pictures are quite extreme and so parents are quite distressed basically so it's putting that particular image into perspective" (Ellen)*

Part of putting information into perspective is also about reassurance, as explained by Joanna;

*"it's just working with the parents just making sure they're aware of that [medical facts] but trying to give them some reassurance that you know two thirds of people with NF1 don't have significant problems"*

These health professionals explained that the variable and unpredictable nature of NF1 makes providing accurate information challenging. Specialists saw themselves as important in patients' lives as they are able to contextualise the information patients have been given.

## **Boundaries**

Health professionals also mentioned areas that were outside of their remit such as psychological support. They described the difficulties involved in explaining their role and in getting specialist support for their patients. Karen explains that;

*"In terms of providing ongoing support around that [appearance] I don't know that that necessarily is within the remit of {my service} so I think that's possibly more sort of, if somebody's having particular concerns and issues ongoing which is affecting how they're living their lives we would then probably be looking to see who we could refer onto for additional support."*

She explains that *"we have quite clear boundaries in terms of what we can offer so we're not therapeutic counsellors, we can help with support we can help with adjustment"*

Participants talked of referrals to support groups *"We can be signposter, that's something we can do....we can act as that sort of signposter" (Karen)*

However participants mentioned that finding further support can be problematic and referral routes can be complicated depending on local pathways.

*"it's difficult for children to access appropriate psychological services" (Ellen)*

*"so often I would be in the situation where I am talking to the GP, primary care, saying I think this person may need some additional therapeutic counselling" (Karen)*

### 6.3.3 Theme 2: Family

Participants discussed the impact of NF1 on families as a whole. They discussed how new cases of NF1 are different to instances where it is inherited, particularly in terms of parents' expectations. Parents who had the condition themselves were thought to have more concerns related to education whilst those for whom the condition was new worried more about appearance and the general prognosis. Participants talked about the concerns that different family members have about the condition, with parents worrying about their children but also children worrying about their own future. There was a lot of discussion about how families are a unit and source of information themselves and how health professionals' role in supporting children can be about supporting parents to support their children. They also discussed how diagnosing a child as having NF1 can give rise to other family members, including a parent, being diagnosed.

#### Inheritance patterns

In discussing the ways people manage NF1 Joanna suggested that it depends on *"who else is affected in the family or not affected, are they the only one"* this was echoed by Karen who, when discussing typical family concerns said *"it varies for every family every individual, it depends on past experience. Is this something in their family that they've known about for many years or is it all completely new?"* She continued *"I think if it's completely new the hardest thing is uncertainty"*.

Ellen explained why parents who have NF1 themselves might react differently to those whose children are the first to be diagnosed *"with the parents of children with NF1, particularly with the mothers who are also affected with NF1, it has had a dramatic effect on their lives and particularly when there is longevity; when grandparents have NF1, parents have NF1 and I've got NF1."* In contrast she explained how in families where the condition is new, *"if there's not a family history and it's a child that is the first in their family with NF1 they're just sort of shell shocked"*.

At different points in the interviews participants discussed feelings of blame, fear and guilt that parents sometimes reported in connection to having NF1 in the family. An example of this was Ellen talking about a child who has been diagnosed with NF1 which the mother did not realise was a pre existing condition in her ex-partner's family; *"mum still can't get over that, she's really very distressed and she was unaware her former partner had a genetic condition"*. Ellen also highlights that *"in a lot of instances they [parents] have a guilt complex because they've given this condition"*.

The uncertainty of the condition and parental concerns for their child's future were seen as central to the general wellbeing of families and regardless of inheritance patterns, health professionals felt that the diagnosis and management of NF1 was often difficult for parents. Joanna mentioned that *"it's a natural parental instinct to want to protect your child and I think anything that you, you fear, and that the parents fears you know thinking ahead for the child"*. Participants were very aware of the need to support all members of the family.

### **Multiplicity of relationships**

Participants reflected that they often supported and worked with whole families and may be treating a person as a patient, and also as a parent of a child with NF1; this could potentially make relationships challenging. Families may have several members with NF1 and the variability of the condition may mean that different family members have very different medical, social and psychological experiences of the condition.

Joanna explained that *“it (NF1) is variable even amongst family members so for instance if I had a family where maybe they had four or five children you might find that three children are affected plus the parent’s affected. All four of them would have different things associated with different issues there to be dealing with and it depends on what those issues are as to whether there would be further follow ups some families just want to get the genetics done get the diagnosis and then sort it out for themselves”*.

Joanne gave an example of a situation when the roles can be confusing *“when you go into schools, I always go in with a parent and very often it’s the parent that’s, if they are affected, that’s the parent that goes in and its yeah it’s an interesting experience talking about the characteristics of someone with NF1, a child with NF1, when the parent is sat there next to you that also has NF1”*.

Ellen discussed how having NF1 in the family can impact across generations and what this can mean for young people, particularly in relation to the learning difficulties that can be associated with the condition. *“They’ve not presented as being significantly delayed so they’ve just been labelled as well borderline and coming from a slow family”*.

Participants described how their role of supporting families as a whole included supporting and advising parents on talking to their child about NF1.

*“We talk to families about how you might start to talk to children about neurofibromatosis”* (Karen)

*“I have found that parents will often ask how do we talk to them about this. What do we say about the marks? What do we say about the lumps? What do we say about this, what do we say about that? My advice to parents is to talk to them at a level that they understand, to answer their questions at a level that they will understand”* (Joanna)

While there was a consensus that information should be given gradually and that parents were often the best people to inform their child about NF1, these health professionals recognised that this was not always successful. As Joanna explains; *“I do feel uncomfortable when the parents say that we won’t tell them just yet. I know certainly there are occasions where if I feel that families are struggling with the diagnosis, I would suggest seeing the family without the child”*

### 6.3.4 Theme 3: Adolescence

Participants were explicitly asked about the medical and psychosocial services they provided, or were aware of, for young people with NF1. There seemed to be a consensus that adolescence was not a time during which patients had a lot of contact with professionals. However, all the participants talked about how difficult adolescence can be for young people with the condition.

#### Invisible

The health professionals discussed having child and adult patients but had little contact with young people. Participants talked about young people as not being engaged in healthcare and mentioned concerns that, during the transition from paediatrics to adult care, some may “*slip through the net*” (Joanna).

*“We tend to see people at the younger age or when they’re adults thinking about families themselves”* (Karen).

Ellen explained how lack of continuity and not being seen, or slipping through the net, can be problematic for these young people. She gave the example of a patient who did not attend clinic appointments during early adolescence and came back on his own accord as a young adult by which time because he had not been for many years, it was deemed too late to surgically manage his plexiforms, which had been distressing for him.

When asked about how young people seek or access support interviewees felt that they did not explicitly ask them for help. Participants mentioned occasionally receiving referrals where 16/17 year olds wanted to know a little more about their condition, but health professionals felt that young people were unlikely to be proactive in contacting them.

#### Hard to reach

This sub theme highlights why adolescents with NF1 may not be engaged in healthcare. Health professionals suggested it may be because parents book appointments and manage healthcare processes when the young person is a child, but when young people do it themselves it can be problematic. In part this was a practical issue relating to some young people being at school or college during the day and finding it difficult to contact the service. Additionally, health professionals wondered if contacting professionals might feel daunting for some young people.

Joanna explains “*I think very few teenagers will actually voluntarily make an appointment with a GP*”.

The practical side was highlighted by Karen who explained she often had to talk to parents and leave messages with them as they are home when she calls, and the young person is at school, college or work, “*The service operates between nine and five and children are often at school at that time I do*

*try and make a conscious effort to book those kind of calls [to young people] at the end of the day so at about four o' clock or something when there's a chance they'll be home from school .."*

Participants recognised the importance of engaging with young people and talked about having an open door policy and being there as children and young people needed them, highlighting the importance of ongoing relationships. *"Building up a good rapport with them is the most important thing and getting them to trust you enough that they will feel that they can talk to you about these things"* (Joanna).

As highlighted in research with other professionals working with young people with genetic conditions (eg Callard et al, 2012), participants in the current study agreed that engaging young people is difficult and time consuming, yet very worthwhile and essential.

#### **6.3.5 Theme 4: Appearance**

Appearance and the noticeability of NF1 were mentioned throughout interviews, while this may in part reflect participants' knowledge of the researchers background in appearance, they were also very clear about the importance of the appearance related aspects of the condition for many of the young people, adults and parents they saw. Health professionals felt that parents voiced concerns regarding their child's appearance more often if NF1 was not a pre existing condition in their family. This contrasts with thoughts about families where NF1 was a pre existing condition where concerns were likely to be around providing support (particularly at school) whilst appearance and the noticeability of NF1 were less likely to be the main concerns:

*"I think that if you get parents that have no experience of it, one of the most common questions I get asked is will it affect the face",* (Joanna).

Health professionals talked of the normality of having appearance concerns during adolescence, as well as the specific concerns of young people with NF1. They also discussed managing an altered appearance both practically and emotionally.

#### **Normative discontent**

Health professionals felt that younger children with NF1 generally feel confident showing their body and will talk about café au laits quite openly. For instance, Joanna mentions that *"most children I think will ask 'What are these? What are these marks?' Some children won't even be bothered they'll just accept that well mum's got a lot of birthmarks and I've got a lot of birthmarks"*.

However, participants mentioned that this changed with age and that there is an expectation that young people may have difficulties with appearance-related issues:

*“that’s a difficult time. All sorts of appearance issues with the media you know you’ve got so much pressure from high gloss magazines.....there’s so much pressure over appearance and so much fitting in the norm that you know the slightest deviation outside that is just very very difficult.”* She further highlights that *“most neurofibromas tend to appear at puberty and that’s just awful.”* (Joanna)

Ellen also talked about appearance norms and concerns *“Everything now is aesthetically based, even within the normal population people change their appearance cosmetically like they change their cars. So if you don’t like your nose, get a new one. If you don’t like your boobs, you get them bigger. So you know, in the normal population, there’s a mindset that you can change and you’ve got to strive for perfection and so anybody who falls below an acceptable norm is even more noticeable and more prone to ridicule”*.

There was a general feeling that while children with NF1 are happy with their appearance, this can change as they grow up and are faced with the onslaught of social pressures and messages about appearance:

*“The most fantastic thing about children with NF1 is that they’ve just got the most this, they ooze with enthusiasm and they desperately want to please everyone and desperately want to get things right and do well and yet you know when they come out into the great wide world all we can think about is how people look and that’s really sad because the personalities are there”* (Joanna).

### **Managing an altered appearance**

The uncertain nature of changes to appearance, particularly changes related to neurofibromas and plexiforms, were reported as possible concerns for young people.

Ellen explained how young people may be *“feeling that they might mutate at any stage to some sort of ghoulish creature with lumps and bumps.”* She discussed how uncertainty is difficult to manage and talked of *“.....Russian roulette syndrome because you know mum might be significantly affected or might not be, your sister might be or might not have a mark on her and you think am I going to be the one that falls?”*

These professionals did not feel there was an obvious link between the noticeability of NF1 and adjustment. Joanna explained that *“I think there’s so many environmental factors that can influence how they cope with it [an altered appearance] and also sometimes somebody can have very high tumour load which means they’ve got lots and lots of neurofibromas and be absolutely fine with it, it’s a case of this is how I am this is how you accept me. Other occasions somebody can have very few neurofibromas but for them that’s a major issue”*.

Joanna also explained that it can be difficult to talk to people about their appearance concerns *“I can almost feel that maybe we’re skirting around something and I would say ‘I get the sense that there’s something that bothering’ you and try and tease it out of them”*.

Overall, health professionals felt young people were generally affected by the uncertainty of the condition and that appearance concerns should be addressed. Participants talked of various different ways in which young people were affected by an altered appearance and how they managed and adapted to change often using a range of ways to conceal areas that caused them concern.

*“...they adapt, you talk to adults that have had it for some time, they’ve adapted to it maybe their arms there aren’t any markings on their arms they might wear short sleeve t-shirts they would never go sleeveless....swimming costumes can be a problem” (Ellen).*

Comparing this to Joanna’s quote in the previous section in which she described children with NF1 as oozing with enthusiasm, it would seem that, as they move from childhood to early adulthood, patients become increasingly aware of their condition and feel less confident about their appearance.

As well as everyday practical concerns, appearance was also discussed in terms of medical and surgical management. For instance, *“some of those lumps can be removed if necessary if they are in places that are causing some irritation or are part unsightly or just they just want to have them removed, some people do come and get them removed on a regular basis.” (Joanna)*

Health professionals also mentioned that whilst young people tended not to engage with medical care, the exception is often when neurofibromas are causing particular issues. Karen pointed out that in these situations *“sometimes we do find ourselves sort of saying ‘well actually the best people to talk to about that are the plastic surgeons or dermatologists’ and we would refer them on appropriately if people have concerns and want to know if they can be removed”*.

Surgically managing the appearance concerns of young people with NF1 also included invasive treatments for stature, leg length, and plexiforms. Ellen expressed concerns that issues need to be picked up early if they are to be treated surgically and notes that *“I think, the cosmetic issues, if we could monitor these children more, see them for a medical every six months and every twelve months.....yes if we see changes soon enough with regards to the prognosis with regards to cosmetic surgery might be better.”*

### **Supporting positive adjustment**

This sub theme relates to the perceived consequences of an altered appearance and other emotional and social issues related to NF1. Participants mentioned that parents had concerns around how others would respond to their child and whilst they noted the different reactions of parents with/without

NF1, a common theme was how parents could help support their child and promote a sense of resilience. For instance Karen highlighted that a common concern was *“how are they going to deal with that as parents? How are they going to help their children?”* and that emotional issues for all families may be around *“anxiety – are they going to mutate as they get older?”*

Karen also felt that families who have NF1 had lowered social expectations and, in terms of relationships, some younger adults felt they had to settle for partners who liked them rather than waiting for someone they really loved *“they’ve felt well if I don’t stick with him I’ll never have a chance at romance”*.

In terms of supporting these needs, participants talked of having tried to run groups and involve people in events, but there was a concern that people with NF1 do not want to use support groups. Ellen suggested this may be as *“there’s still there’s a bit of stigma to it they want to know everything about it but they don’t want to be part of a group where there’s other children with NF.”* Joanna suggested that it is important that young people are not lumped together as one group *“they’re not necessarily a group of people, they’re little groups within a big band and I think we don’t always address that sort of thing.”* She also suggested that *“there is a value in commonality of experience and young people being able to come together”*.

#### 6.4 Discussion

The findings presented in the preceding sections provide a descriptive account of the insights and experience of three health professionals who work with young people with NF1 and their families. Participants highlighted the importance of being a contact point and a provider of information for many different groups; parents, children, young people, adults and other professionals (including other health professionals and teachers) and discussed the limits, or boundaries of their role. As previously highlighted, health professionals are particularly important in supporting families’ informational (Metcalf, 2011) as well as educational needs (Suris, 2004). Supporting families in this way, holistically and across ages and generations, was recognised as important by the participants in this study. They described managing different relationships within families and felt inheritance patterns were important in terms of patients’ styles of coping and the concerns reported.

The findings demonstrate participants’ recognition that NF1 can have a psychosocial impact on young people, and their families. However they describe this impact as complicated and, in line with the appearance literature on the nature of severity of visible difference (eg Ong, 2007; Moss, 2005), highlight that noticeability and tumor load are not necessarily indicative of psychosocial adjustment. Overall, participants felt that appearance concerns and strategies for managing an altered appearance were commonly discussed by patients. Furthermore they also highlighted the difficulty in managing appearance changes and uncertainty during adolescence for young people with NF1 and also for the general population. Participants felt that the heightened awareness of appearance during

adolescence is compounded by media focus and socio cultural pressures regarding perfection and idealised images. Previous research reflects this in reports of high levels of appearance-related anxiety amongst adolescents with and without visible differences (Rumsey & Harcourt, 2012).

In line with previous research with people with NF1 (for example Ferner et al, 2007; Ablon, 1999) these health professionals highlighted that challenges, for both parents and young people with NF1, can relate to the uncertainty of the condition as well as its impact on appearance. They felt that it may be the uncertainty of appearance changes that is most worrying for young people, rather than actual changes.

However, whilst health professionals felt that appearance concerns and dissatisfaction may be 'normal' during adolescence, and they could discuss some concerns with patients, they also felt that some young people with NF1 would benefit from additional psychosocial support around managing an altered appearance, or the potential for an altered appearance. In particular, alongside professional psychological input and counselling, health professionals suggested that meeting others with the same condition or having general social support and advice may be beneficial.

A finding that may indicate particular challenges for all those affected by NF1 or working with people with the condition is that these health professionals described this patient population as often having little contact with other health professionals. In considering the possible reasons for this, participants discussed practical difficulties associated with engaging young people in health care and explained that young people are not often a group likely to independently or proactively seek advice, information, or support. Non compliance and non adherence to medical regimens and appointments are well reported amongst young people with chronic health conditions (eg Michaud, et al, 2004). However, adolescence is a time during which health behaviours are often consolidated and adjustment to chronic illness is established (Holmbeck, 2002) and this possible lack of engagement could potentially mean that young people with NF1 are at risk of poor adjustment to and management of their condition.

Health professionals in this exploratory study felt their role was concerned with being a regular point of contact and with providing and contextualising information. They described families who had previous experience of NF1 and those to whom it was a new condition as presenting with different concerns, and worked with whole family units in a holistic manner. Young people were described as difficult to engage and a group that health professionals were not always in regular contact with. With regards to appearance; adolescence was seen as a time when appearance was a concern for many and whilst professionals described some concerns that young people and families had regarding appearance changes and NF1, they also described positive adaptation to a changing appearance.

In conclusion, whilst health professionals discussed different areas of experience, there were three key aspects derived from the themes and subthemes in this section that were central to discussions of young people's experience of NF1 and in need of further exploration.

- First, a key aspect across themes related to appearance concerns and how young people manage an altered appearance. Whilst highlighting the importance of noticeability, health professionals also indicated that tumor load is not indicative of an individual's level of appearance concern.
- Second, the support needs of young people (and their families) were discussed in terms of support groups and information needs, as well as professional psychological support, both in terms of general and appearance-specific support.
- Third, engaging specifically with young people, both in practical terms and in terms of ensuring service provision is 'young people friendly', was discussed as important.

These three areas, (see figure 9) were used as the basis for developing a survey for health professionals. This development and the choice of questions to investigate each identified area are discussed in Chapter 7.

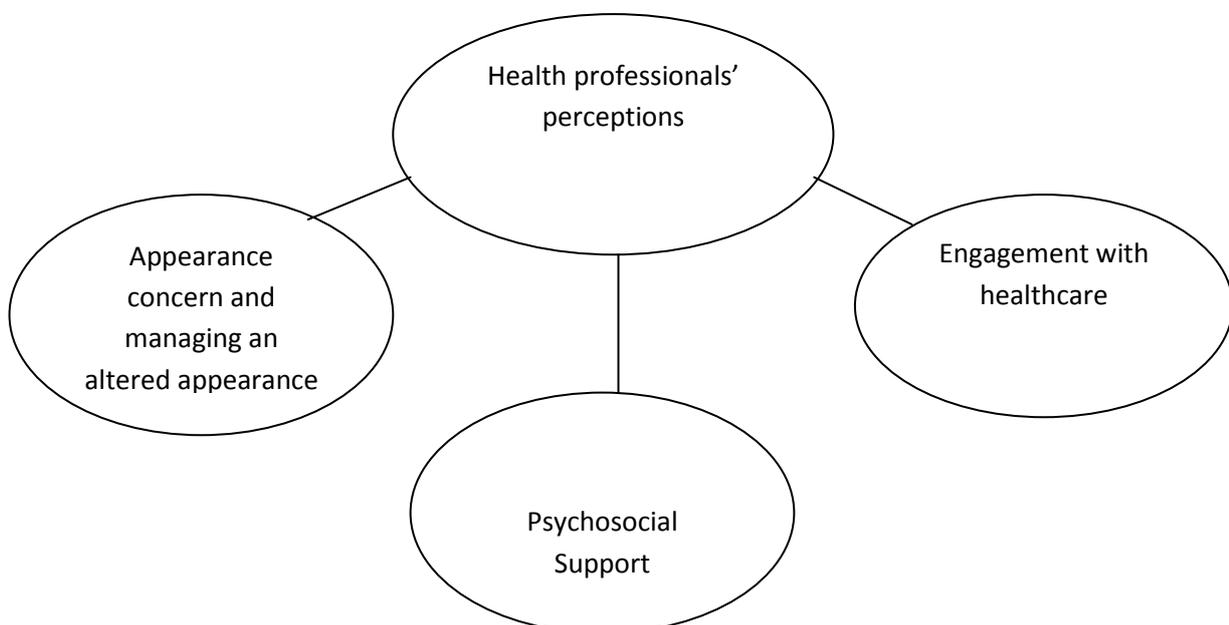


Figure 9: Key aspects of health professionals' perceptions of young people's experiences of having NF1 and their own experience of working with parents, and individuals with NF1

## Interview Studies: Conclusion

The previous three chapters have explored the role of appearance within the broader context of young people's experiences of NF1. How these findings were used to develop surveys is outlined, in detail, in the following chapter. An in-depth discussion of the role that appearance plays within young people's psychosocial experience of living with NF1, as well as how findings from the thesis as a whole relate to the existing literature and the implications for research and care is provided in Chapter 9. The current section provides a commentary on the main similarities and differences found when considering findings from across the three interview studies.

As discussed in Chapter 3, interview guides were developed in order to explore experience of NF1 in broad terms, and to focus on appearance within this. This acknowledged that the impact of the appearance-related aspects of NF1 is likely to be complex. The picture that has emerged is both comprehensive and detailed. At times this entailed discussion in interviews of areas that are not obviously directly related to appearance, however this has enabled participants to discuss the complex way in which aspects related to appearance impact on young people with NF1.

While some aspects of interviewees' experiences overlap, and there were certainly commonalities across the interviews, the three groups also define their experiences and understandings in very different ways. All participant groups highlighted the importance of appearance within the day-to-day management of NF1, in terms of actual changes to appearance, managing uncertainty and managing the reactions of other people. The importance of families and social support was also emphasized by all groups, as were individual perceptions of NF1. Aspects that differed between groups were the importance of noticeability and inheritance patterns as well as aspects of understanding NF1 and engagement with support and healthcare.

A particular concern raised by health professionals related to young people's levels of engagement in healthcare. Parents had similar concerns, primarily in terms of transferring responsibility for healthcare to their child. Parents of young people in adult care (up to the age of 24) discussed feeling a need to check and make sure their child was remembering to arrange appointments. It could be argued that those taking part in an interview of this sort are likely to be highly motivated and engaged with issues associated with their NF1, and the young people in this study did appear to be very engaged in their healthcare. Some felt they had become more responsible for it as they became older, but younger participants also described high levels of engagement and were aware of when checkups should be arranged, which hospitals, departments and professionals they were under the care of, and what symptoms to watch for and be aware of. Young people perceived health professionals as having a lack of knowledge of NF1 and this was identified as a possible challenge to young peoples' engagement. Young people made a distinction between specialist and non specialist health professionals and it was important to young people and parents that health professionals understood them and were knowledgeable about NF1.

Understanding and managing NF1 was also discussed by each participant group. Parents discussed diagnosis being a time of information seeking and learning about NF1. Young people generally felt that learning about NF1 either happened gradually or they talked of reconceptualising the condition as they grew up. Young people wanted information specifically designed for them, and a chance to talk about their condition. This was also highlighted by health professionals and parents; there was awareness amongst all participant groups that information and support, specific to young people, is important to their understanding of and adjustment to NF1.

Whilst all groups felt that managing an altered appearance as a result of NF1 could be challenging, particularly in terms of managing social situations and others' responses, the way in which it was discussed differed a great deal. Young people primarily discussed appearance in terms of their concerns about possible future changes. Whilst some discussed current concerns, for many the uncertain nature of their appearance was the most challenging aspect of NF1. Parents generally discussed their child's appearance in terms of how noticeable the visible aspects of NF1 were, including the site of tumors or other visible differences. Health professionals discussed how patients managed appearance concerns and the changing nature of concerns through childhood into adulthood. They felt that children were generally positive about their appearance whilst young people and adults were more worried about the impact of NF1 on appearance. Health professionals described a growing awareness of appearance amongst young people and described how people learnt strategies and ways of managing any appearance-related concerns as they grew up.

Health professionals reported conversations with parents and young people who they felt may benefit from better social skills and from meeting others with the same condition. Managing social situations and others' responses to their appearance was central to young people's experiences. Parents also highlighted this as a key issue. The participant groups differed however in terms of the importance of the noticeability of NF1 within this social experience. Some young people reflected that even if other people could not see a mark or a scar, they themselves knew it was there and that this made them feel awkward. Health professionals expressed views similar to young people and highlighted that the number of tumors or the visibility of NF1 was not necessarily indicative of positive or negative feelings towards appearance and concerns about social interactions. Parents however felt that how other people reacted to their child's visible difference was related to how visible any difference was. The impact of the perceived noticeability of NF1 is an area that will be investigated further in the next stage of this research.

Some families had members with NF1 before the young person was diagnosed, some were diagnosed at the same time as their child and, for others, NF1 was very new to them and the young person was the only family member to be diagnosed with the condition. However, while this information was provided as a direct answer to a question within interviews, it did not seem to be relevant within or between experiences. During analysis there did not seem to be particular differences in the way in which young people and parents discussed their experience when taking

inheritance patterns into consideration. However, health professionals described inheritance patterns as important in determining how families (and young people) managed their condition and its impact. This will be explored further in questionnaires.

The first stage of this research has provided a rich description of young people's experiences of NF1 from the perspectives of young people themselves, parents and the health professionals who treat them. Within this some young people and parents described NF1 as having little emotional impact and explained it was a small part of their lives but others described it as difficult and challenging and something they worried about daily. Health professionals also reflected that the impact of NF1 varies greatly between different individuals and families. Across interviews the role of appearance within young people's experiences of NF1 was clearly an important factor. This thesis now moves on to identify specific areas to be investigated in surveys of young people, parents and health professionals. The following chapter demonstrates how this was accomplished whilst a detailed consideration of this process and the strengths and limitations of the methods used in this thesis, is provided in Chapter 9.

## Chapter 7: Survey Development

This chapter considers the aim of this research phase and the research questions guiding the survey development. It then describes, in detail, how the findings presented in the previous chapters were used to develop three surveys.

### 7.1 Moving from stage 1 to stage 2

Chapters 4, 5 and 6 each concluded by presenting the key aspects that each group (young people, parents and health professionals) felt were important to young people with NF1 (see figure 10).

<b>Young people</b>	<b>Parents</b>	<b>Health professionals</b>
(a) general feelings about appearance	(a) child's management of social situations and others' responses	(a) appearance concern and managing an altered appearance
(b) social situations and others' responses	(b) parents' coping strategies and perceptions of NF1	(b) psychosocial support
(c) perceptions of NF1	(c) appearance and noticeability of their child's NF1	(c) engagement with healthcare

Figure 10: Key aspects of experience/perceptions of young people, parents and health professionals identified in interview studies

Moving from the first stage of this research (interviews) into the second (surveys) involved unpicking these key aspects and focusing on the factors within them that related to the role of appearance within young people's experience and which could be explored further. This in turn led to the formation of a series of research questions which defined the focus of this research stage (see section 7.2). The next task was to consider how the research questions could be answered. This entailed revisiting the literature reviewed previously and, in some cases, exploring new areas of literature. This process led to the identification of standardised measures and, where such measures were not appropriate, questions using quotes from the previous interviews and open ended questions were used to explore specific aspects further. As discussed previously in Chapter 3, Creswell et al (2003) and Bryman (2008) both underline the importance of being explicit in showing how methods are mixed within a mixed methods project. With this in mind, a description of this process is provided in the following sections.

Before moving on to outline research questions and describe each survey design, it is important to reflect that moving from one research stage to another was challenging. There were many aspects of young people's experiences that would have been interesting to take forward, and leaving areas behind was particularly difficult. For instance many interesting points were made in interviews about school and healthcare experiences that would have been worthwhile to pursue further. However, this thesis does not aim to measure and explore all aspects of young people's experiences, but rather explored experiences broadly in interviews in order to focus on how and where appearance played a role. Moving from interview analysis to survey development entailed focusing on these specific aspects, how this was accomplished and the choices made are detailed in the following sections.

## **7.2 Research aims and questions**

The aim of the second stage of this programme of research was to explore the role of appearance within specific aspects of young people's experience of NF1, quantifying and comparing findings within and between participant groups. The following research questions were developed from the findings described in the previous stage:

### ***Young people:***

- How do young people with NF1 feel about their appearance in general and do they report their condition as noticeable?
- How do young people report their social comfort and interactions with others and is this different for those who report their NF1 as noticeable or not?
- What are young people's perceptions of NF1 and do they differ for those who report NF1 as noticeable or not?
- How do perceptions of NF1, general feelings about their appearance, subjective noticeability, social comfort and interactions with others impact on young people's wellbeing?
- How do perceptions of NF1, general feelings about their appearance, social comfort and interactions with others relate to one another?
- Do young people's reports of appearance-related concerns, social comfort, interactions with others, perceptions of NF1 and general wellbeing differ according to demographic factors?
- How do young people describe their information and support needs and how is appearance mentioned within this?

### ***Parents:***

- How do parents describe the role of appearance within their child's experience of NF1 and how noticeable do they feel their child's condition is?

- How do parents report their child's social comfort and interactions with others and does this relate to parents reports of noticeability?
- How do parents report that they cope with their child having NF1? Are there specific ways in which parents cope and do coping patterns relate to whether or not the condition was inherited?
- What are parents' perceptions of NF1 and do these relate to the perceived noticeability of their child's condition?
- How do parents perceptions of NF1 relate to their views of their child's social comfort and interactions with others?
- How do parents describe their (and their child's) information and support needs and how is appearance mentioned within this?

#### ***Young people and parents:***

- How do parents and young people differ on their descriptions of social comfort, interactions with others and perceptions of NF1?

#### ***Health Professionals:***

- How do health professionals describe NF1 impacting on young people and how is the role of appearance described within this?
- What psychosocial support do health professionals feel would help young people with NF1 to adapt positively to NF1 and the appearance related aspects of the condition?
- How do health professionals describe their role in young people's management of NF1?

### **7.3 Survey Design**

This section concentrates on describing how the findings presented in the previous three chapters have informed the development of surveys for young people (aged 14-24) with NF1, parents of young people aged 14-24 with NF1 and health professionals who work with people with NF1 in order to answer the questions outlined above.

#### **7.3.1 The young peoples' survey design**

Having defined the areas to explore further in the survey, a decision was made to develop questions using quotes from interviews and asking participants to agree/disagree as well as open ended questions and standardised measures thus allowing for quantification of existing findings, the possibility of new findings and the comparison of data with other studies that have used the same measures.

### 7.3.1.1 Demographic information

Young people were asked for demographic information (including gender, age and ethnicity) and were asked to confirm that they had NF1 and whether anyone else in their family had the condition. These questions were asked in order to describe the participant group as a whole and also so findings could be explored using these factors.

As discussed previously (see Chapter 3), a particular consideration when developing interview guides was not to introduce areas of concern to young people. Interviews highlighted the variability in young people's knowledge of NF1 and its genetic nature. While a few participants discussed inheritance and the possibility of future children having the condition, others did not seem to be aware of this. However, some young people did comment on the closeness of relationships with other family members with NF1 and health professionals explicitly stated in interviews that there was a difference between families where NF1 was a pre existing condition and those where it was new. With this in mind, a decision was made that the young person's survey would ask 'Does anyone else in the family have NF1?' and if they answered 'yes' respondents were asked what relation. Whilst it is acknowledged that this may not have provided accurate inheritance information, it was used to explore if there were differences between those who knew whether they did or did not have other family members with the condition.

A second area that needed careful consideration was whether or not to ask young people about learning and behavioural difficulties. When developing the interview guide a decision was made not to include questions on this topic, but rather to discuss experience generally, including school and then decide during analysis of interviews and the development of the survey whether learning difficulties should be explored in the second phase (see section 3.4). As discussed in previous chapters, the possible impact of learning and behavioural difficulties on young people's experiences is highly complex. Specifically, strong social skills are identified as important in mediating the challenges of an altered appearance and therefore, if young people with NF1 have specific difficulties that impact on social skills and behaviour, this could be important within their management of the appearance related aspects of the condition. Interviews highlighted the diverse and complex nature of discussing learning and behavioural difficulties; in particular some participants were unsure if they had a specific diagnosis. Any impact discussed generally related to experience at school including frustrations linked to having needs recognised and met. A decision was made that exploring exactly if and how learning difficulties impacted on young people's experiences of the appearance related aspects of NF1 was not within the remit of this thesis. However as these topics were important to some young people, the survey included questions about school experience. This gave young people the opportunity to express the importance of learning and behavioural difficulties should they wish, but equally young people could indicate that NF1 did not make any difference at school. An open ended question asked for 'any other comments about school', this was included in order to explore whether appearance was a particular concern in school or if there were other areas that young people felt were important.

### 7.3.1.2 Appearance

In discussing the overall impact of NF1, across interviews participants discussed feelings about their appearance in many different ways; they discussed NF1 specific concerns such as plexiforms and neurofibromas, they talked of feeling self-conscious and covering up signs of the condition. They also talked generally about feeling anxious or aware of the way they looked and discussed concerns that their appearance might change in the future. Some discussed the noticeability of their condition as a worry, others said noticeability could be positive as other people were more likely to be supportive. Some were more concerned about non NF1 related aspects of their appearance. Overall, whilst some young people were positive about their appearance and felt NF1 had little impact, others found the visible signs of NF1, or the possibility of visible signs, very challenging. In order to explore this further in the survey, young people were asked how they feel about their appearance in general terms using the Body Esteem (BE) Scale (appearance subscale Mendelson et al, 2001), they were also asked whether they felt their NF1 was noticeable to others (yes or no) and to identify their main concerns and support needs in open ended questions. The aim of these questions was to identify how young people with NF1 feel about their appearance, whether perceived noticeability impacts on this and if they identify appearance as a main concern and support need. These areas are detailed below.

Mendelson (2001) defines body esteem as the self-evaluation of one's own appearance; it is a person's attitude to their own body. The BE appearance subscale measures young people's overall feelings about their appearance; it does not refer specifically to size or weight but rather examines how happy people are with the way they look. The appearance sub scale of the Body Esteem Scale for Adolescents & Adults (Mendelson, 2001) was chosen as it relates to general feelings about appearance and has consistently predicted self-esteem independent of weight and attributions of others. Mendelson et al (2001) suggest the clinical implication of the scale is that the best route to self-esteem is through improving general feelings about appearance. The Body Esteem scale was developed with a sample of 1334 young people aged 12-25 (Mendelson, 2001) and has been widely used internationally with clinical and non-clinical populations including young people with chronic conditions and in general population groups with adolescents and young adults (Pinquart, 2013; Forbes et al, 2012; Streeter et al, 2012; McVey et al, 2003). Furthermore the measure has been previously used with young people with a visible difference (Lawrence et al, 2007). The BE-appearance subscale consists of 10 items. Respondents are asked to indicate how often they agree with a series of statements about their appearance such as *'I like what I see when I look in the mirror'*, *'I wish I looked like someone else'* and *'I worry about the way I look'* on a 5 point scale ranging from *'Never'* (0) to *'Always'* (4). The body esteem appearance sub scale has good internal consistency with cronbach alpha coefficient reported of .92 (Mendelson, 1997). In the current study the cronbach alpha coefficient was .9.

During interviews participants discussed the noticeability of their NF1 in many different ways. In several cases it seemed that those least visibly affected were the most concerned about their appearance. As previously discussed in Chapter 2 there is evidence to suggest that subjective rather than objective measures of appearance and disfigurement predict wellbeing (eg Moss, 2005; Ong et al, 2007). As objectively measured severity of a visible difference does not consistently relate to individual adjustment, previous research with people with a visible difference (including Rumsey et al, 2010) has measured subjective rather than objective accounts of severity. A relationship between subjective severity and adjustment has been demonstrated by Moss and Carr (2004) and Rumsey et al (2004). Furthermore Barton and North (2007) suggest future studies should ask young people to rate the severity of their NF1 themselves, rather than rely on objective third party assessment. Therefore, young people taking part in the survey were asked if their NF1 was noticeable to other people (yes or no) and, if so, how. This was then used to investigate if people who consider their NF1 noticeable to others feel differently about their appearance, social experience and perceptions overall.

In addition the survey asked young people to complete open ended questions, '*For me NF1 is.....*' and '*My main concern about NF1 is.....*' One aim of these questions was to investigate whether appearance was identified as a main concern and whether descriptions of NF1 included appearance related aspects. These open ended questions allowed for the possibility of new findings to be raised in this stage of the research programme and were used to triangulate earlier findings from interviews with young people that highlighted appearance as an important concern.

### **7.3.1.3 Social situations and others' responses**

Consistent with the literature outlined in Chapter 2, the second key aspect identified related to young people's social comfort and managing the responses of others. These experiences were examined in the survey using the Perceived Stigmatization Questionnaire (PSQ), and the Social Comfort Questionnaire (SCQ) developed by Lawrence et al, (2010; 2006b).

The Perceived Stigmatization Questionnaire (PSQ), measures the frequency of various stigmatising social behaviours experienced by people with visible differences. It has 21 items and asks "*during your normal day you probably see and talk to many different people. We want to know how often people act in certain ways towards you. For each question rate how often people do certain things. Make your ratings about how people treated you over the last year.*" Responses are marked on a 5 point scale ranging from 'Never' (1) to 'Always' (5). Items include statements such as '*People are friendly with me*' and '*People pick on me*'. The PSQ is comprised of three sub scales; (1) absence of friendly behaviour, (2) confused/staring behaviour and (3) hostile behaviour as well as a total perceived stigma score. Scores are calculated by reverse coding items on the absence of friendly behaviour sub scale and dividing each subscale total by the number of items in the subscale, the total PSQ score is produced by adding together all item scores and dividing by the total number of items.

Scale scores and total scores are then averages in the same metric as the 5 point frequency scale on the questionnaire (Never, Almost Never, Sometimes, Often, Always).

The Social Comfort Questionnaire (SCQ) measures social isolation and the violation of privacy for people with a visible difference. Violation of privacy refers to increased staring and questions being asked about the appearance which was highlighted as a particularly challenging aspect of their experience of NF1 by some participants during interviews. The scale has 8 items and asks respondents to indicate (again on a 5 point scale) how often they feel or think a series of statements including *'I feel like I fit in with most groups'* and *'I like meeting new people'*. Scores are calculated by reverse coding three items, then adding together all items and dividing by the total number of items. The final score is the average of items; it is on the same metric scale as the 5 point frequency ratings used for questions (Never, Almost Never, Sometimes, Often, Always).

The PSQ and SCQ have been validated with adults, (Lawrence et al, 2006b) and children and young people (Lawrence et al, 2010). Scales have good internal consistency with cronbach alpha coefficient reported with adults of .93 for overall stigmatisation, .91 for absence of friendly behaviour, .92 on confused/staring behaviour and .88 for hostile behaviour while .91 is reported for the SCQ (Lawrence et al, 2006b). In the current study, the cronbach alpha coefficients also demonstrated good internal consistency; .91 for overall stigmatisation, .78 on absence of friendly behaviour, .89 on confused/staring behaviour, .88 for hostile behaviour and the SCQ internal consistency was .87. The PSQ and SCQ have been used with adults and children with burns (Lawrence et al, 2010; 2007; 2006 a; 2006 b).

#### **7.3.1.4 Perceptions of NF1**

In line with previous literature (eg Ferner et al, 2007), interviews suggested that NF1 is a highly varied and unpredictable condition. Analysis of the interviews highlighted the variability of the young people's perceptions of NF1 and the impact they felt it had on their lives. Young people discussed many different physical symptoms as well as different understandings of what the condition was and described how these perceptions changed over time. For instance as outlined in Chapter 4, Sarah described feeling NF1 was a 'leg thing' as a child and went on to explain how she had reconceptualised the condition and now understood it as a genetic condition. Like many others, including Daniel, she explained that there was a growing awareness through adolescence into adulthood of the genetic nature of the condition, its possible impact on future children and possible complications of the condition including its impact on her appearance. People with NF1 may face challenges in conceptualising their condition due to the general lack of a coherent representation of NF1. Ablon (1999, p8), writing about the impact of NF1, suggests *"...the cultural context and state of professional and lay understandings about a condition are significant factors in shaping person's remembrances and recounting of events. The image an affected person has of his/her illness and future is closely tied to their perceptions of their doctor's, family's, friend's and society's views of their*

*condition*". Interview findings suggest that young people hold many different perceptions of what NF1 is and how it may affect them, formed through their contact with health professionals, the way in which their parents talked about NF1 and the information and support they had received. The survey investigated these perceptions of NF1 in order to explore the role of appearance within perceptions and to examine whether there was a relationship between how people perceive NF1 and how they feel about their appearance and social experience.

When people have a condition, such as NF1, they hold a set of perceptions about it which are important in determining how they feel about and manage their condition. They are dynamic and changeable, responding to an individual's ideas about their condition and are influential in the individual's emotional response and coping behaviours. These perceptions come from many different avenues and have been found to impact on psychosocial outcomes. This is described within the Common-Sense Model (CSM) of Illness Representations (Leventhal et al., 1980; see figure 11).

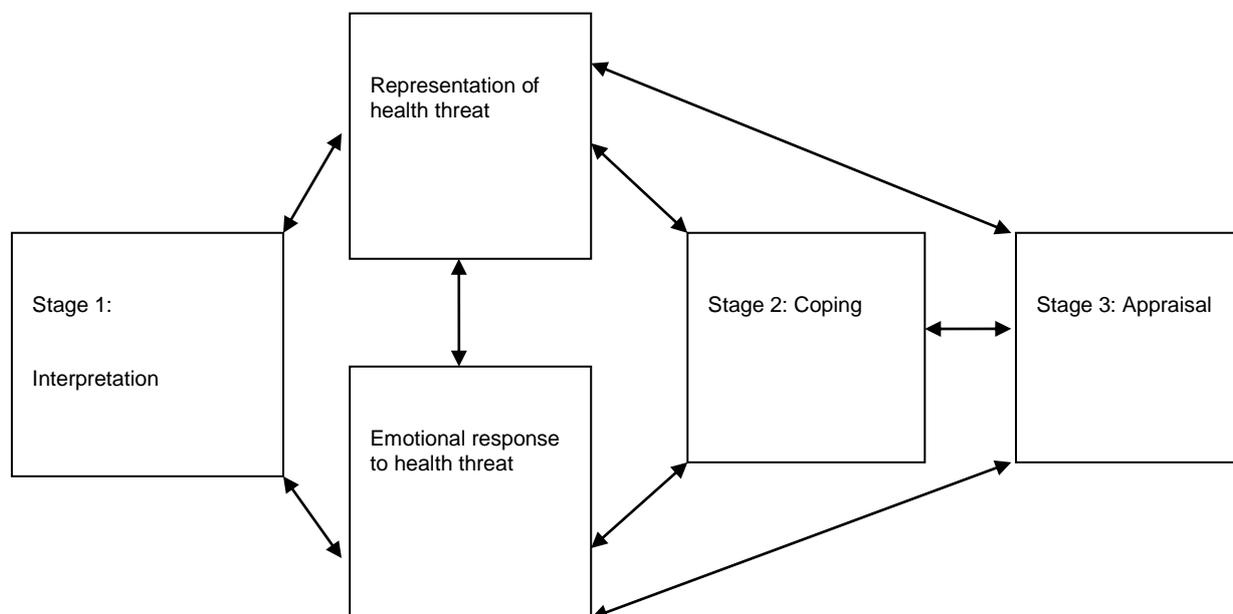


Figure 11: Leventhal's self-regulatory model of illness behaviour

This model proposes that illness perceptions give people a framework for coping with and understanding their illness and suggests these perceptions develop over time, and from different avenues including interactions with health professionals, medical experience and treatment, family communication, type of information received as well as previous experience with the condition. Leventhal et al (1980) identified five cognitive dimensions which comprise illness perceptions; identity (label or medical diagnosis and symptoms experienced), perceived cause (biological, psychosocial or both), timeline (beliefs about how long illness will last), consequences (individual's perception of the effect of the illness on their life), and curability/controllability (whether or not illness can be treated and cured and how controllable the illness is). Alongside the cognitive dimensions, Leventhal et al (1980) proposed that how people feel about their condition emotionally impacts on the formation of

perceptions. Cognitive dimensions and emotional response form a crucial part of Leventhal's self-regulatory model of illness cognitions which assumes that given a problem or challenge individuals are motivated to solve the problem and re-establish normality. This generally involves three stages. The first stage is interpretation and making sense of the problem, then using strategies to attempt to manage or cope with the problem and finally appraisal of the coping activity. Illness perceptions are a crucial part of this process at all three stages.

The five cognitive dimensions can be measured using the Illness Perceptions Questionnaire (IPQ, Weinman et al. 1996). The later revised version, IPQ-R (Moss-Morris et al, 2002) incorporates measurement of illness coherence (how individuals understand their illness) and emotional representation, reflecting the emotional response component within Leventhal's original model (see figure 11). The IPQ-R asks respondents to rate a series of statements about their illness on a scale of 0-10. This questionnaire has been used with people with different health conditions and ages. The IPQ-R has over 80 items and as such was determined to be over long for use in this instance. Therefore in order to investigate illness perceptions in the current study, the Brief IPQ (B-IPQ) was included in the questionnaire. The B-IPQ is based on the IPQ-R and was validated by Broadbent et al (2006) who found it to be a valid and reliable measure of illness perceptions in a variety of illness groups. The B-IPQ has been widely used with adult patient groups (including palliative care, diabetes, arthritis, bipolar disorder), children (Chong et al, 2010) and young people with cancer (Jamison et al, 1986), diabetes (Edgar & Skinner, 2003) and cystic fibrosis (Bucks et al, 2009).

The B-IPQ is comprised of 9 items, each assessing a dimension of illness perception; 5 assess cognitive perceptions (consequence, timeline, personal control, treatment control, identity) and 2 assess emotional representations (concern and emotions). One item assesses illness coherence (understanding). These items are each measured on a scale of 0-10. The score suggests how benign or threatening an illness is with a high score suggesting a threatening view of the condition. The Causal item (item 9) is assessed with an open ended question which asks what the respondent considers to be the most important causes of the condition. This item is grouped into categories and reported. In this study, the word 'illness' on the B-IPQ, as suggested by Broadbent et al, was replaced by 'NF1'.

In order to examine how the items on the B-IPQ may help provide an understanding of young people's perceptions of NF1 they were considered alongside evidence from the interviews with young people, as shown in table 3.

<b>Dimension on B-IPQ</b>	<b>Definition</b> <i>(Broadbent et al 2006)</i>	<b>Examples from interview data</b>
Identity	Label or medical diagnosis and symptoms experienced	While NF1 is the overall diagnosis, symptoms were described as diverse, uncertain and changeable. People are treated in different clinics for different presenting symptoms. When asked about their condition, participants described it in many ways including 'a leg thing', 'learning difficulties' 'neurofibromas/plexiforms' and 'ADHD' suggesting NF1 may not have a clear identity, an important part of this identify was often physical aspects such as cafe au laits and neurofibromas.
Cause	Can be biological, psychosocial, both or other.	The genetic nature of the condition means inheritance is either from a parent or de novo; some participants spoke of luck and chance and some had lay theories about inheritance (including alcohol consumption)
Timeline	Beliefs about how long illness will last- acute/chronic/cyclical	Participants displayed uncertainty around this. Certain physical symptoms are seen as acute (ie some neurofibromas change shape/size and can be removed) whilst the underlying condition is chronic. It is important to recognise that with this condition the timeline may extend past an individual, i.e. it may have been inherited from a parent and can be passed onto a child.
Consequences	Patient's perception of the effect of the illness on their life-physical, emotional and both.	There was a great deal of uncertainty and concern around both the physical and emotional effects of the condition. People had specific appearance related concerns and a more general consequence was living with the uncertainty of the condition (which itself often related to the uncertainty of possible appearance changes).
Control (personal & treatment)	Whether or not illness can be treated and cured and how controllable the illness is either by themselves or others (such as doctor).	Some people felt that whilst the condition itself cannot be cured, physical symptoms may be controlled; this could be medically or through information seeking or homeopathy. Some expressed concerns that non-specialist health professionals knew little about the condition. Participants described feeling they had little or no control over their NF1, in particular the lack of control over their physical appearance was described as challenging by some participants.

Coherence	Understanding of the illness	Young people's understanding of NF1 was found to be important within their accounts during interviews, while some young people were very knowledgeable about their condition, others described feeling unclear about what NF1 was and how it might progress. Participants described their understanding of NF1 changing during adolescence.
Concern and emotion	Relate to the emotional impact of the condition	Interview findings indicated that young people with NF1 may have a range of diverse emotional responses. Some participants described great concern and worry while others described NF1 as having little impact and was just a part of their life.

Table 3: Examples of Leventhal et al's cognitive dimensions within young people's interview data

The B-IPQ shows reliability and concurrent validity with existing measures of illness perceptions and correlations with relevant subscales of the IPQ-R are highly significant (ranging from 0.32 to 0.63, Broadbent et al). In the current study the overall cronbach alpha coefficient was 0.23 which is a little low, which may be in part due to the item on treatment being difficult for people with NF1 to answer since there is no cure or treatment for the condition overall although treatment is available for symptoms. It would appear from comments on questionnaires that some participants interpreted this question as treatment for NF1 (ie a cure) while others related this to treating symptoms. If this item were deleted the cronbach alpha coefficient would be 0.7. The B-IPQ was used to measure how individuals perceive NF1 and specifically how this relates to other aspects of their experience (overall wellbeing, body esteem and social experience).

Young people's perceptions of NF1 are likely to have been formed through interactions with medical professionals, their experiences at school and the way in which the condition is portrayed in the media as well as their support and information needs being met. These are vast areas to explore in depth, yet it was considered important to explore areas that young people had identified in interviews as significant within their understanding of NF1 and its general portrayal in order to consider whether these impacted on the role of appearance. Questions exploring specific aspects of school, healthcare, media, information and support were developed in three ways; first using quotes from interviews which respondents were asked to agree or disagree with such as, '*the internet is a good place to find information on NF1*'. Second presenting multiple choice questions based on interview data '*Where do or did you find information about NF1*' and third respondents were asked for any further comments regarding healthcare, school, support and information.

### 7.3.1.5 General wellbeing

The survey study explored how the variables identified thus far relate to one another and impact on young people's general wellbeing. In order to assess this, a measure of general wellbeing was used. This involved a consideration of what is meant by wellbeing and how it can be measured. In discussing the impact of NF1, several young people had said they were very happy and just got on with life and they talked about the importance of 'seeing the positives'. Happiness is essentially a way of explaining subjective well being, it relates to a positive sense of fulfilment, contentment, enjoyment of life and pleasure, and is often seen as one of the most important goals for life (Veenhoven, 2013; Diener & Oishi, 2000). It has been linked with many measures of positive wellbeing and is associated with many benefits across life. In a meta analysis, Lyubomirsky et al (2005) suggest that happiness may be both a consequence of different successes and a cause, and they provide evidence highlighting the link between happiness and many positive outcomes in work, relationships and health. The relationship between happiness and health is well documented (Borghesi & Vercelli, 2010); the World Health Organisation emphasises happiness as a component of health (DeGargino, 2004) and governments (including the UK) have begun to measure it as a measure of national progress (Bulletin of the World Health Organization, 2011).

In order to assess young people's happiness the Subjective Happiness Scale (SHS) (Lyubomirsky & Lepper, 1999) was included in the survey. The SHS is based upon the evidence that objective circumstances, demographics and dispositional factors are not strongly correlated with happiness or positive wellbeing. People can consider themselves happy in spite of personal circumstances that would seem to predict otherwise. In order to examine both positive and negative experiences and to investigate how some young people have positive outcomes, while others find living with NF1 more challenging, associations between the SHS and other measures of appearance satisfaction, illness perceptions, stigma and social comfort will be explored. The SHS has been used in studies with children, young people and adults (Holder et al, 2012; Moghnie & Kazarian, 2012; Swami, 2008). It is a four item scale of global subjective happiness each using a seven point response scale. According to Lyubomirsky & Lepper (1999) the SHS has good internal consistency with cronbach alpha coefficient reported ranging between .79 and .94. In the current study the cronbach alpha coefficient was .88.

As mentioned previously, the survey also asked young people to complete two open ended questions, 'For me NF1 is.....' and 'My main concern about NF1 is.....'. While these were included primarily to explore whether participants cited appearance related concerns, these two questions were also included in order to give young people the opportunity to express what NF1 means to them and how it impacts on their life and general wellbeing.

A table detailing the use of themes in developing questionnaires can be found in appendix 17 and the questionnaire for young people (including measures as used in the questionnaire) can be found in appendix 18.

### **7.3.2 The parents' survey design**

The process of developing a survey to explore the key aspects and research questions identified earlier is detailed in the following sections.

#### **7.3.2.1 Demographic information**

Respondents' gender, ethnicity, the age and gender of their child who has NF1 was sought and respondents were asked to confirm their child had NF1, if they had NF1 themselves and also whether the condition was inherited or new to the family. These questions were asked in order to describe the participant group as a whole and also so that findings could be explored using these factors.

#### **7.3.2.2 Child's management of social situations and others' responses**

Many parents described feeling concerned about how other people reacted to and treated their child; some were worried about their child's social life and social skills; some discussed being proactive and attempting to ensure their child had positive social experiences. In order to examine parents' perceptions of their child's social experiences and any stigma experienced, the Perceived Stigma Questionnaire and Social Comfort Questionnaire were employed. The PSQ and SCQ were used in the young people's survey and are outlined in detail in section 7.3.1.3. The questions differed in asking parents to consider statements in relation to their observations of their child's experience and how their child thinks and feels, statements on the PSQ included '*people are friendly with my child*' and '*people bully my child*' while statements on the SCQ included '*he/she feels that no one understands them*'. As outlined previously responses are made on a 5 point scale (never to always). Using the questionnaires in both the young peoples' and parents' surveys also enabled a comparison of how the two groups reported these experiences.

As previously reported (Section 7.3.1.3) the PSQ and SCQ scales have good internal consistency; the scales have been validated with child and adult burn survivors (Lawrence et al, 2010; 2006b). The PSQ has been used previously to compare parent and child perceptions of stigma, again with burn survivors (Lawrence et al, 2011) and with children/adolescents with acquired and congenital facial differences and their parents (Masnari, 2012). The SCQ has not been used in this way previously. For this study, parents cronbach alpha scores on the PSQ are reported as .89 on the PSQ overall and .82 to .86 on subscales. In the current study the cronbach alpha coefficients demonstrate good internal consistency; .94 for overall stigmatization, .88 on absence of friendly behaviour, .91 for confused/staring behaviour, .93 for hostile behaviour and the SCQ internal consistency is .90.

### 7.3.2.3 Coping

Managing the impact of NF1, including the appearance related aspects of the condition was discussed throughout interviews. In general terms parents talked of their lives changing and shifting and described a period of adjustment to the condition followed by long term changes to family life. Parents discussed managing the appearance related aspects of NF1 in terms of worrying about how their child felt about their appearance and how others responded to their child. The ways in which parents manage the challenges they and their child might face can be seen as coping strategies, defined by Folkman et al. (1986) as a person's constantly changing cognitive and behavioural efforts to manage specific taxing external and/or internal demands. Research suggests there may be a relationship between a child's wellbeing and parental coping strategies (Cavallo, 2009), meaning that targeting parental coping strategies and supporting the development of positive strategies may be an important way in which health professionals can support family adaptation to a chronic illness (McCubbin & McCubbin, 1988).

However, it is important to recognise that coping is not a clear concept and there is much ongoing debate within health psychology regarding both the concept itself and its measurement (Lazarus 2000) and the body of literature in this area has been much criticised (Somerfield & McCrae, 2000). As Folkman and Moskowitz (2004) stress "*Coping is not a stand-alone phenomenon. It is embedded in a complex, dynamic stress process that involves the person, the environment, and the relationship between them*" (p. 748). Identifying how people manage challenge is important in explaining why and how some individuals manage stress well and others do not. Understanding this process in greater detail has the potential to inform care and interventions to support effective ways of managing challenge. Exploring how parents cope with having a child with NF1 and identifying positive strategies that support wellbeing may be highly supportive of both parents' and young people's experiences. However, this could be worthy of a PhD in its own right. Therefore, in recognition of the complexity of coping as a concept, and in order to ensure a clear focus for exploration, parental coping was examined within the survey in precise terms. Specifically, the coping strategies that parents used were investigated in order to examine first, if any particular strategies were common to families with a child with NF1; secondly if parents' reports of the noticeability of their child's NF1 were related to specific coping strategies. Finally coping was examined in order to explore if there were, as suggested by health professionals, differences in coping patterns between those who reported that NF1 was a pre existing condition in the family and those for whom their child was the first to have the condition.

The Coping Health Inventory for Parents (CHIP) (McCubbin et al, 1983) was designed to assess appraisal of the behaviours parents use to manage family life when they have a child with a chronic illness. The CHIP is comprised of 45 behaviours grouped into three patterns: pattern one focuses on behaviours that strengthen family life and relationships and parents outlook on life/optimism, pattern two focuses on parents' efforts to develop relationships with others and engage in activities that

enhance their individual identity and self worth as well as behaviours that manage tension and pressure, pattern three concentrates on parents' relationships with healthcare professionals and other parents of chronically ill children. This includes knowledge and understanding of the illness.

The scale items are different coping behaviours; respondents indicate if the behaviour is not helpful (0), minimally helpful (1), moderately helpful (2) or extremely helpful (3). If respondents did not use the behaviour they can record this by ticking 'chose not to use it' or 'not possible'. The CHIP is scored by adding together the total scores in each area (total scores can therefore range from 0-135, while pattern one ranges from 0-57, pattern 2 0-54 and pattern 3 0-24). The scale has been widely used over many years with many different chronic health conditions including with parents of children with diabetes and asthma (Holden et al, 1997) and Juvenile Idiopathic Arthritis (Cavello et al, 2009). According to McCubbin, et al the three subscales of the CHIP have good internal consistency with a Cronbach alpha coefficient reported of 0.79 for subscale one (family life and optimism), 0.79 for subscale two (maintaining own wellbeing) and 0.71 for subscale three (relationships with health professionals and other parents with chronically ill children). In the current study the Cronbach alpha coefficient for the subscales were 0.87, 0.87 and 0.82 respectively.

#### 7.3.2.4 Perceptions of NF1

Like young people, parents reported many different perceptions of NF1, both in terms of how it affected them as a parent and how it affected their child. To explore both of these aspects further and compare them to other measures in the survey, the Brief IPQ was used in the parental questionnaire to describe and measure parents' illness perceptions of NF1. This measure is discussed in detail in section 7.3.1.4. To assess parents' perceptions of the effect of NF1 on both themselves and their child additional questions were included, specifically:

- How much does NF1 affect your child's life? *and* How much does NF1 affect your life?
- How well do you feel you understand NF1? *and* How well do you feel your child understands NF1?
- How much does NF1 affect you emotionally? *and* How much does NF1 affect your child emotionally?

Two overall B-IPQ scores were then calculated, one reflected the perceived impact of their child's NF1 to the parent and one reflected the perceived impact to their child. In the current study the cronbach alpha coefficient was .62 for perceived threat to child and .64 for perceived threat to self.

Parents and young peoples' scores on the B-IPQ items were compared in order to investigate if perceptions differ between these groups. Previous research has identified differences in illness perceptions (measured with the IPQ-R) between mothers and their children (aged 11-17 years) with diabetes (Olsen et al, 2008). Furthermore van der Velde et al (2011) found a significant difference in

adolescent and parent illness perceptions (using the B-IPQ) of food allergy, on the item 'illness concern' and 'emotional representation' (parents reported greater emotion and concern than young people).

Like young people, in interviews parents discussed the many different ways in which their perceptions of NF1 may have been influenced for example via their child's experiences at school, healthcare and the information and support available for them and their child. As in the young persons' survey, these were explored further using quotes from parents' interviews and asking participants the extent to which they agreed or disagreed, as well as multiple choice questions and comment boxes.

### **7.3.2.5 Appearance and Noticeability**

In interviews, parents expressed different views on the impact of the appearance-related aspects of NF1 and appearance in general. Some described the importance of appearance changes through adolescence and others felt their child did not have any particular concerns. In order to explore parents' perceptions of the role of appearance they were asked how often their child expressed concern about their appearance (generally and NF1 specifically), and how confident they, as a parent, felt managing concerns about appearance.

The role of appearance across their experiences as a parent of a child with NF1, as well as their perceptions of their child's experience, was explored by asking, in an open ended question, how NF1 affects (a) them and (b) their child. Additionally, a feature of parents' accounts in interviews was that concerns related to NF1 appeared to change over time. Therefore this was explored by asking parents what their concerns were at initial diagnosis and at the time of completing the questionnaire.

Parents were also asked about the noticeability of their child's NF1, in terms of how noticeable they thought it was to others on a scale of 0 (not at all) to 10 (highly noticeable). This differed from how young people themselves were asked about the noticeability of their NF1 (The young people's survey asked 'Do you think your NF1 is noticeable to other people?' with the response options 'Yes' or 'No'). In part this difference resulted from a concern that asking young people to rate how noticeable their NF1 was on a scale seemed unethical. But it also seemed that parents' discussions of the noticeability of NF1 were different to the accounts of young people themselves. Young people discussed concerns related to the noticeability of NF1 in two main ways. First they discussed whether others could notice their NF1 or not. Second young people discussed the unpredictability of NF1 and their concerns about potential changes and the possibility of greater noticeability in the future. Parents however, discussed being aware of and sensitive to other people's reactions to the noticeability of their child's condition. Parents particularly talked about the importance of the extent of noticeability and the merits of symptoms being visible on different sites on the body, for instance several parents discussed how neurofibromas on the torso were easier to manage than ones on the face. Interview

findings suggest that for parents the extent of the noticeability of their child's NF1 was an important factor in their assessment of their child's experiences.

A table detailing the use of themes in developing questionnaires can be found in appendix 19 and the questionnaire for parents (including measures as used in the questionnaire) can be found in appendix 20.

### **7.3.3 The health professionals' survey design**

This section outlines how the key aspects identified from the analysis of interviews with health professionals and the subsequent research questions were used to develop a survey of health professionals who work with young people with NF1 (including genetic counsellors, nursing staff, geneticists, dermatologists and paediatricians). While the previous two surveys used the analysis of qualitative data to develop a questionnaire to further examine direct psychological experience, this current survey aims to investigate health professionals' views and perceptions of young people's and parent's experiences. As such the questionnaire for health professionals did not include standardised measures but rather used open ended questions and quotes from interviews, seeking to quantify the commonality of viewpoints and experiences within a larger and more diverse population. The way in which the aspects informed the development of the questionnaire is detailed in the following sections.

#### **7.3.3.1 Demographic information**

Health professionals were asked for their job title and how often they saw patients with NF1. A particular point raised in interviews by professionals was a view that the impact of NF1 varied by inheritance patterns. In order to examine this further, the questionnaire asked participants for the most common concerns of families where the condition is (a) inherited and (b) de novo.

#### **7.3.3.2 Appearance**

The questionnaire presented a series of quotes from interviews with health professionals regarding young people's appearance-related concerns and respondents were asked the extent to which they agreed or disagreed. Questions related to how children and young people feel about the way they look, whether this changes and then, if so, how.

#### **7.3.3.3 Psychosocial support**

This was examined firstly through asking participants to identify the most common concern for young people with NF1 in a text box. This question was open ended in order to allow for new ideas and considerations. Across interviews health professionals discussed the support that is or should be

available for young people with NF1. The questionnaire examined this further by asking what support professionals considered was and should be available.

#### **7.3.3.4 Engagement with healthcare**

Professionals' roles and the different relationships they develop and manage with young people and parents were evident across the interviews with health professionals. Questions examined these relationships and asked if and how supporting parents and families is part of the professional's role and whether parents require help and support in talking to their children about NF1. Questions examined health professionals' opinions of young people's and their wider family's management and engagement in healthcare. Health professionals were asked to identify how often their role involves a series of activities that had been mentioned across interviews including offering support at initial diagnosis, and providing psychosocial and practical support for young people and families, being a point of contact and signposting to other support services.

A table detailing the use of themes in developing questionnaires can be found in appendix 21 and the questionnaire for health professionals can be found in appendix 22.

This chapter has described how findings from the qualitative stage of this research were used in order to develop questionnaires for young people, parents and health professionals. The following chapter moves on to describe how the survey study was carried out and presents findings from the three surveys.

## Chapter 8 Survey findings

The current chapter describes ethical considerations and approvals, the recruitment of participants and data collection followed by four results sections; findings from the young people's survey, the parents' survey, a comparison of data from measures used in both parents' and young people's surveys, and finally the health professionals' survey. An in-depth discussion of the role that appearance plays within young people's psychosocial experience of living with NF1 is provided in Chapter 9 which discusses and evaluates the findings from the thesis as a whole.

### 8.1 Ethical approval and considerations

The study was approved by the Ethics Committee of the Faculty of Health and life Sciences at The University of the West of England. In addition, as the young people's surveys and parents' surveys were sent to participants through NHS clinic lists, ethical approval was obtained from NRES committee South West (Frenchay) and further R&D approvals were sought and granted at each individual clinic site.

Each questionnaire was available online using a web based survey site, Qualtrics, and also as a paper-based version. The online and paper questionnaires had the same content, the only difference related to consent. The return and completion of the paper questionnaire was taken as consent to take part, the online questionnaire asked respondents to tick a box to indicate they had read the information and consented to take part. Before completing the paper copy those under 16 years were asked to provide a parent/carers signature to confirm the parent/carers had read the information sheet and was happy for their child to take part. Any questionnaires returned without parental consent were not included in the data analysis. The online questionnaire asked respondents under 16 years to discuss the questionnaire with their parents/carers and to provide a parent/carers email. Once the questionnaire was completed the parent/carers was sent an email explaining their son/daughter had taken part in a study. A blank questionnaire and information sheets were attached to the email along with the researcher's contact details. Parents/carers were asked to reply to the email saying they were happy for their son/daughter to take part. Any participant whose parent/carers did not reply was not included in data analysis and an email was sent to the parent explaining this.

The language used in the questionnaire was carefully examined, in order to ensure that it was accessible for all potential participants. Two children under 14 years (the minimum age included in the survey) read the information sheets and completed the questionnaire and information (online and paper) and were asked to comment on any words or phrases they did not understand. In addition the draft questionnaires were sent to all participants from the interview study and comments were invited. Feedback was positive; and only minor changes were made to the information sheet.

## 8.2 Recruitment, procedure and sample size

The inclusion criteria was:

- (a) Young people aged 14-24 years (inclusive) with a diagnosis of NF1
- (b) Parents of young people who were aged 14-24 years with a diagnosis of NF1 (parents were not excluded for having a diagnosis of NF1 themselves but it was not an inclusion criteria)
- (c) Health Professionals who work with young people/patients with NF1

Participants were recruited internationally (as discussed in Chapter 3) and had to be able to complete a questionnaire in English, in order to take part.

A power analysis identified that a minimum of 30 young people and 30 parents would be needed to have at least 80% power in detecting a statistically significant relationship at standard levels of significance ( $\alpha = 0.05$ ; two-sided). Similar sample sizes would apply to measures of correlation when investigating the strength of the relationship within parent and young person couples. Caution dictates that the sample sizes needed to be inflated to approximately  $n = 40$  to retain similar power if nonparametric statistics are needed. With this in mind, the aim was to recruit 40 participants for each of the three surveys.

A range of recruitment strategies were employed. With regards to the health professionals' survey, those who were aware of the research were asked to circulate the survey to their colleagues. NF1 specialist clinics and professionals who specialise in NF1 were then identified by searching the internet for regional genetics clinics listings and specialist services and clinics. Details of the questionnaires were emailed directly to individuals where email addresses were freely available online, and to admin and generic hospital addresses if not (see appendix 26 for copy of email). Additionally the researcher attended an annual meeting of UK based genetic counsellors/nurses in order to promote the study and questionnaires were given out to delegates at the event; a list of parties who were interested in receiving further information to forward to colleagues was compiled and these individuals were emailed with survey details. Finally professional organisations relating to genetics and NF1 in the UK, Ireland, USA, Canada, New Zealand and Australia were contacted. The following associations agreed to forward details to their membership, through newsletters and/or email distribution lists; British Society for Genetic Medicine (BSGM), Association of Genetic Nurses and Counsellors (UK), Canadian Association of Genetic Counsellors (Canada), Australasian Society of Genetic Counsellors (ASGC, Australia and New Zealand) and Genetic Alliance UK (for example see appendix 25)

Young people and parents were recruited through several avenues. Firstly information was sent to young people and their parents from clinics at three NHS sites in England; Central Manchester University Hospitals Trust (NF specialist centre), Oxford University Hospitals NHS Trust (Genetics

department) and Great Ormond Street Hospital (Genetics department of a specialist children's hospital). Staff members at Central Manchester University Hospital were aware of and supportive of this research from the very start and as such were approached to take part in the study and were asked for advice on who else may be interested in taking part. It was on this basis that Oxford University Hospital was approached. Finally, a particular objective was to recruit young people from across the defined age range (14-24), both Oxford and Manchester clinic lists would include young people across this range. However recruiting younger participants was more challenging in the interview stage. A decision was made therefore to approach a children's hospital, specifically to take part as their clinic list would be comprised of a younger age group.

The researcher visited each hospital and with the support of local staff accessed clinic databases and patient notes in order to obtain the details of young people who fit the study inclusion criteria. A note was left in each patient's record explaining that they had been contacted regarding the study. Envelopes were then addressed and packs posted onsite. The study packs included a cover letter from their consultant, a postcard with the web address of the online version of the survey and questionnaires for young people and their parents (see appendix 18, 20 & 27). The letter explicitly stated that both young people and parents/carers did not have to take part. Packs were addressed to young people aged 16 or over and to the parent/carer of young people aged 14 or 15. Those wishing to participate could either complete the enclosed questionnaire or could complete the survey online. In addition, details of the survey were included on web sites, internet forums and newsletters of the Centre for Appearance Research, Changing Faces (UK), The Neuro Foundation (UK), The Childrens Tumor Foundation (USA), British Columbia Neurofibromatosis Foundation (Canada), and Neurofibromatosis Midatlantic (USA) (for examples see appendix 23 & 24). In addition a research facebook page and profile was created and details of the study were posted on various facebook pages relating to NF1 (with the page owner's permission). Finally some members of social media sites chose to publicise the research themselves, by sharing details either on their own facebook page, or in other online groups.

Young people and parent/carers surveys were sent out and available online between May and September 2012. Surveys for health professionals were provided between April and August 2011. Survey data was analysed between September and November 2012 (see appendix 18, 20 & 22 for copies of the surveys and information packs)

### **8.3 Data Analysis**

The frequencies and mean scores of non-standardised quantitative questions were calculated and presented. The quantitative data resulting from standardised measures was analysed using the statistical program SPSS (version 19), after checking the distribution of variables, examining histograms and checking for outliers by examining boxplots. The use of parametric tests was found to be justified.

Qualitative data was analysed using content analysis. The aim of content analysis is that text is classified into smaller categories that can be quantified. Content analysis aims to look for meaning across a text in a systematic manner (Elo & Kyngas, 2007; Morgan, 1993). Stemler (2001) writes that the main benefit of the method is that it is systematic and replicable and can deal with large volumes of data. Stemler cautions that the method should go beyond a word count and that categories should be well defined and mutually exclusive. In the survey analysis, open ended responses to questions were compiled into a list and were read several times, initial codes were identified and all data was then coded into this list. Data was then quantified by counting the frequency of each code.

#### 8.4 Young people's survey findings

Seventy six young people completed the questionnaire. Of these, three were excluded from data analysis as they were under 16 and parental consent was not received. As such 73 questionnaires were included in analysis (22 paper copies of the 180 sent out through clinic lists and 51 which were completed online). All participants confirmed that they had a diagnosis of NF1, 63% (n=46) were diagnosed before they were five years old, 12.3% (n=9) between five and ten years, 16.4% (n=12) aged 11-16 and 2.7% (n=2) aged over 18. One person was unsure and a further three participants did not answer. 34.2% of participants had a family member with NF1. Of these, 84% said a parent had NF1. 58.9% of participants (n=43) had no family history of it and 6.8% (n=5) were unsure. Further demographic information is provided in table 4:

<b>Gender</b>	Female	n=52 (71.2%)
	Male	n=20 (27.4%)
	Not provided	n=1 (1.4%)
<b>Age</b> mean=20.4 (sd 2.91)	14	n=2 (2.8%)
	15	n=2 (2.8%)
	16	n=4 (5.6%)
	17	n=7 (9.9%)
	18	n=6 (8.5%)
	19	n=3 (4.2%)
	20	n=8 (11.3%)
	21	n=11 (15.5%)
	22	n=5 (7%)
	23	n=10 (14.1%)
24	n=10 (18.3%)	
<b>Ethnicity</b>	White	n=59 (80.8%)
	Mixed	n=6 (8.2%)
	Asian	n=5 (6.8%)
	Black	n=2 (2.7%)
	Not provided	n=1 (1.4%)

<b>Education and Employment</b>	Full time education	n=38 (52.1%)
	Part time study & work	n=5 (6.8%)
	Full/part time work	n=12 (16.4%)
	Unemployed	n=15 (20.5%)
	Stay at home parent	n=1 (1.4%)
	Not provided	n=2 (2.9%)
<b>Country of residence</b>	England	n=39 (54%)
	Scotland, Wales, N Ireland and Ireland	n=9 (12%)
	North America	n=16 (22%)
	Other (Europe, New Zealand, Australia, Philippines, South America & China)	n=8 (11%)
	Not provided	n=1 (1.4%)

Table 4: Surveys of young people: Respondent demographic details (NB, due to rounding not all tables add up to 100%)

The table below provides descriptive statistics for the standardised measures used in this survey. Findings are then discussed in relation to the research questions outlined in section 7.2.

<b>Scale</b>	<b>N</b>	<b>Min</b>	<b>Max</b>	<b>Mean</b>	<b>Possible Range</b>	<b>Std. Dev</b>
Subjective Happiness Scale (higher score indicates greater happiness)	69	4	27	18.19	4-28	5.465
Brief-Illness Perceptions Questionnaire (higher score indicates more threatening view of NF1)	69	20	75	48.30	0-80	11.532
PSQ subscale1 (absence of friendly behaviour) (Higher scores indicates higher levels of perceived stigma)	61	1	4	2.46	1-5	.556
PSQ subscale 2 (Confused/staring behaviour) (Higher scores indicates higher levels of perceived stigma)	64	1	4	1.98	1-5	.811
PSQ subscale 3 (Hostile Behaviour) (Higher scores indicates higher levels of perceived stigma)	63	1	4	2.08	1-5	.867
Perceived Stigma Questionnaire (Higher scores indicates higher levels of perceived stigma)	58	1	3	2.19	1-5	.585
Social Comfort Questionnaire (higher scores indicate higher levels of social comfort)	65	1	5	3.10	1-5	.778
Body Esteem (appearance subscale) (Higher scores indicate greater body esteem)	68	0	4	2.01	0-4	1.126

Table 5: Descriptive scores on the standardised measures used in the survey of young people

### How do young people with NF1 feel about their appearance in general and do they report their condition as noticeable?

General feelings about appearance were measured by the body esteem appearance subscale. Body esteem scores are calculated on a scale of 0-4 with higher numbers indicating greater body esteem. Mean body esteem scores in the current study ( $M=2.01$ ,  $SD=1.13$ ) were slightly lower than published means for similarly aged participants (Mendelson et al, 2001). Lawrence et al (2007) suggest scores lower than one indicate very low body esteem, adopting this cut off then 25% ( $n=17$ ) of young people in this study scored in this range while 33.9% ( $n=23$ ) scored three or four indicating positive body esteem.

Participants were directly asked if they thought their NF1 was noticeable to others (yes or no) and 47.1% ( $n=33$ ) of participants felt it was while 52.9% ( $n=37$ ) felt it was not. An independent T-test was used to explore if there was a significant difference in scores on the BE (appearance subscale) for those who reported their NF1 as noticeable or not noticeable. There was no significant difference in scores for participants who reported NF1 as noticeable ( $m=1.91$   $SD=1.254$ ) and those who reported it was not noticeable ( $m=2.14$ ,  $SD=1.004$ ;  $t(65) = 0.856$ ,  $p=0.395$ , two tailed). The magnitude of the differences in the means (mean difference= $0.237$ , 95% CI:  $-0.315$  to  $0.789$ ) was small.

Participants were asked in an open ended question for their main concern about NF1. The aim of this question was to explore how often appearance was cited as a main concern. Sixty four participants responded. Using content analysis, responses were coded into eight categories as shown in Table 6. Specific medical concerns were reported most frequently, these related to pain, treatment and ongoing issues. The second most common concerns were the uncertainty of appearance related aspects of NF1. Concerns relating to having children, were cited by almost a quarter of participants. While a few people reported specific appearance related concerns appearance was more often discussed alongside concern for the future. The uncertain nature of NF1 and concerns relating to possible future changes to their appearance appeared to be concerning in terms of both possible medical and appearance changes.

Code	% and number of responses	Example of quotes from open question
present appearance concern	8% ( $n=5$ )	<i>'it affects the way I look'</i> (Respondent 63)
specific medical concern	28% ( $n=18$ )	<i>"my scoliosis"</i> (Respondent 26)
appearance changes in the future	25%, ( $n=16$ )	<i>'that one day I will be completely covered in bumps and I won't be able to be seen in public'</i> (Respondent 18)
having children	23% ( $n=5$ )	<i>"Having children and being a bad mother by passing on my faulty gene to my child"</i> (Respondent 11).

Learning difficulties and educational issues	6% (n=4)	<i>"that my learning disabilities don't hold me behind when I go to college"</i> (Respondent 32)
social concerns	6% (n=4)	<i>"How I will be affected by it socially"</i> (Respondent 41)
others not knowing about NF1	2% (n=1)	<i>"that someone who should know what NF is does not"</i> (Respondent 64)
No Concern	2% (n=1)	<i>"I have no concerns"</i> (Respondent 76)

Table 6: Main concern: A content analysis of young people's survey responses

Respondents were also asked to complete the sentence 'For me NF1 is.....' in whatever way best described their experience of NF1. Again this question aimed to explore whether people would highlight appearance as an important aspect of NF1. Sixty eight people responded to this question. Within these responses, appearance was not often expressly mentioned and when it was this was in differing ways. Three codes were identified as shown in table 7.

<b>Code</b>	<b>% and number of responses</b>	<b>Example of Quotes from open question</b>
<b>Challenges</b>	44.3% (n=30)	<i>'For me NF1 is like a curse'</i> (respondent 47)  <i>'.....a burden that I feel very lonely carrying'</i> (respondent 11)
<b>Finding positive outcomes</b>	37.7% (n=26)	<i>'Challenging at times, but it has changed my life for the better. I see things differently. NF1 has shaped me to be a stronger person and more understanding'</i> (respondent 22)  <i>'..... what makes me different from the rest of my folks. I have learnt to embrace it... and it doesn't matter what people think of me'</i> (respondent 52)
<b>NF1 is not something that bothers me</b>	18% (n=12)	<i>' part of my life and it doesn't bother me'</i> (respondent 76)  <i>'something that explains who I am. Not ashamed of it and it makes me want others to know about it.'</i> (respondent 43)

Table 7: What is NF1? A content analysis of young people's survey responses

**How do young people report their social comfort and interactions with others and is this different for those who report their NF1 as noticeable or not?**

Analysis of interviews with young people identified social comfort and perceived stigma as important aspects of their experience of NF1. These were examined in the survey using the PSQ and SCQ (Lawrence et al, 2006b). High scores indicate greater perceived stigma and social comfort (see descriptive statistics in table 5). Participants perceived the most stigma relating to the absence of friendly behaviour ( $m = 2.46$   $SD .556$ ). Overall, scores suggest low to moderate levels of perceived stigma, 36.2% ( $n=21$ ) of participants reported perceived stigma in the 'sometimes' range and 63.8% ( $n=37$ ) scored in the range of 'never' to 'almost never'. No participants scored total perceived stigma in the 'often' or 'always' categories. The social comfort mean score ( $m 3.10$   $SD .778$ ) indicates that participants felt moderately socially comfortable. The majority of participants (84.6%,  $n=55$ ) scored social comfort in the 'sometimes' and 'often' range, 13.8% ( $n=9$ ) reported low levels of social comfort and 1.5% ( $n=1$ ) felt social comfort 'always'.

An independent T-test was used to explore if there was a significant difference in scores on the PSQ and SCQ regarding reports of noticeability. There was no significant difference in scores on the SCQ for participants who reported NF1 as noticeable ( $M=3.13$   $SD=.763$ ) and those who reported it was not noticeable ( $m=3.07$ ,  $SD=.814$ ;  $t(62) = -0.297$ ,  $p=0.767$ , two tailed). The magnitude of the differences in the means (mean difference=-0.059, 95% CI: -0.453 to 0.336) was small. There was also no significant difference in scores on the PSQ for participants who reported NF1 as noticeable ( $M=2.26$   $SD=.654$ ) and those who reported it was not noticeable ( $m=2.13$ ,  $SD=.520$ ;  $t(55) = -0.820$ ,  $p=0.416$ , two tailed). The magnitude of the differences in the means (mean difference=-0.129, 95% CI: -0.443 to 0.186) was small.

**What are young people's perceptions of NF1 and do they differ for those who report NF1 as noticeable or not?**

Perceptions of NF1 were explored quantitatively with the B-IPQ. Findings related to each item are presented in this section and are discussed in the following chapter. Analysis demonstrated that NF1 had a moderate impact on young people's lives (consequence  $m 5.33$   $SD 2.81$ ) but scores were widely distributed and 27.4% ( $n=19$ ) of participants felt it had a severe affect on their life (defined as scoring 8-10). The high score on timeline ( $m 9.44$   $SD 1.48$ ) could be expected for a lifelong genetic condition such as NF1 (five participants actually commented that this question was pointless). Participants feel little control over their condition, particularly in terms of personal control ( $m 3.34$   $SD 3.09$ ) with 29% of participants ( $n=21$ ) scoring this at 0 (absolutely no control). Treatment control scored higher ( $m 4.13$   $SD 2.70$ ) although 12.7% ( $n=9$ ) scored this at 0. NF1 symptoms are highly varied and a mean score of 5.04 ( $SD 2.88$ ) on the identity item is a result of scores distributed across the scale. Concern about NF1 scored a moderate 6.22 ( $SD 2.94$ ), however again scores were distributed across the scale and this included 8.2% ( $n=6$ ) scoring 0 and 17.8%  $n=13$  scoring 10)

suggesting concern was highly variable. Understanding of NF1 had a mean score of 6.53 (SD 2.56) however, again scores were wide-ranging suggesting varied understanding. Emotional representation had a mean score of 6.01 (SD 3.21) including 19.4% (n=14) scoring 10 (extremely affected emotionally) and overall 52.7% (n=38) scored 7 and above. The causal items on the B-IPQ were coded into categories; each category was cited by at least three participants. The most frequently cited cause of NF1 was genetics.

An independent t-test comparing the B-IPQ scores on each item for participants according to self reported noticeability of NF1 found no significant differences no significant difference in scores on the B-IPQ for participants who reported NF1 as noticeable (M=49 SD=.12.24) and those who reported it was not noticeable (m=47.44, SD=.11.26;  $t(65) = -0.564, p=0.575$ , two tailed). The magnitude of the differences in the means (mean difference=-1.619, 95% CI: -7.355 to 4.116) was small.

### **How do perceptions of NF1, general feelings about their appearance, subjective noticeability, social comfort and interactions with others impact on young people's wellbeing?**

Young people's general wellbeing was assessed using the subjective happiness scale (descriptive data in table 5). Scores (out of 7, with higher scores indicating greater subjective happiness) indicated young people with NF1 are moderately happy (M=4.72, SD 1.40). 61% rated themselves as slightly to extremely happy, 17.3% were slightly to extremely unhappy while 21.7% scored in the neutral range. Scores are within the range (4.63–5.07) reported by Lyubomirsky and Lepper (1999) for high school and college students (mean age 19 years) in the United States.

An independent T-test was used to explore if there was a significant difference in scores on the SHS for those who reported their NF1 as noticeable or not noticeable. No significant difference was found in scores on the SHS for participants who reported NF1 as noticeable (M=4.7 SD=.1.473) and those who reported it was not noticeable (m=4.73, SD=1.427;  $t(65) = -0.01, p=.992$ , two tailed). The magnitude of the differences in the means (mean difference=-0.004, 95% CI: -0.706 to 0.699) was small.

A multiple regression analysis was then employed in order to explore whether the variance in happiness scores can be explained by scores on other measures. The predictor variables were the BE (appearance), SCQ, PSQ and B-IPQ with happiness as the outcome measure. Preliminary analyses were conducted to ensure no violation of the assumptions of normality, linearity, multicollinearity and homoscedasticity.

The variance explained by the model as a whole was 49%,  $F(4, 50) = 12.077, p.001$ . Only the BE (app) scale was statistically significant (beta = .479,  $p<.005$ ) suggesting that the BE appearance subscale explains almost half of the variance in happiness.

### How do perceptions of NF1, general feelings about their appearance, social comfort and interactions with others relate to one another?

The relationship between the B-IPQ, SHS, PSQ, SCQ and BE (appearance) was investigated using Pearson product moment correlation coefficient (see table 8). Preliminary analysis was performed to ensure no violation of the assumptions of normality, linearity and homoscedasticity.

Higher scores on the B-IPQ indicate a higher perceived threat from the illness. It correlated negatively with happiness (medium effect) social comfort (medium effect) and body esteem (large effect) and positively with perceived stigma (medium effect).

Higher SHS scores indicate greater subjective happiness; this correlated positively with social comfort (large effect) and negatively with B-IPQ (medium effect), PSQ (medium effect) and body esteem (large effect).

Higher PSQ scores indicate higher levels of perceived stigma. This correlates positively with B-IPQ (medium effect) and negatively with happiness (medium effect) body esteem (large effect) and SCQ (large effect).

Higher social comfort correlates with happiness (large effect) and body esteem (large effect) and negatively with B-IPQ (medium effect) and PSQ (large effect).

Finally there is a positive correlation between body esteem and happiness (large effect) and social comfort (large effect) and a negative correlation with B-IPQ (large effect) and PSQ (large effect).

Scale	1 Brief-IPQ	2 Happiness	3 PSQ	4 SCQ	5 BE (app.)
1 Brief-IPQ	-	-0.440**	0.323*	-0.413**	-0.519**
2 Happiness		-	-0.485**	0.529**	0.667**
3 PSQ			-	-0.673**	-0.559**
4 SCQ				-	0.29535**
5 BE (app)					-

\*\* . Correlation is significant at the 0.01 level (2-tailed).

\* . Correlation is significant at the 0.05 level (2-tailed).

Table 8: Survey of young people: Correlations between standardised measures

### Do young people's reports of appearance-related concerns, social comfort, interactions with others, perceptions of NF1 and general wellbeing differ according to demographic factors?

The impact of (a) gender and (b) whether or not other family members had NF1 was investigated across all measures using t-tests. No significant difference were found on any measure between

those who identified as male or female and those who indicated they did have a family member with NF1 and those who did not (See appendix 28 for details of t- tests related to gender, other family members). The associations between participant's age and measures used in the survey were investigated using Pearson product moment correlation coefficient. Participant's age was not significantly associated with any of the measures used.

### **How do young people describe their information and support needs and how is appearance mentioned within this?**

In order to explore support and information needs, participants were asked about their experiences with schools, health professionals, as well as representations of NF1 in the media and avenues of support and information. This data is presented here and is discussed, in terms of how this may inform individual perceptions of NF1 and provide a greater understanding of the role of appearance within perceptions, in the following chapter.

Nine percent agreed that "most people have heard of NF1" and 26.5% (n=17) agreed that "TV magazines and the internet show realistic examples of NF1". The majority (82.6%, n=52) agreed that the internet is a good place to find information on NF1. The most common sources of support were family (64%, n=47) and friends (59%, n=43) followed by boyfriend/girlfriend (23% n=17) and the internet (23%, n=17). Opportunities to meet other young people with either NF1 or other chronic health conditions/learning difficulties and online support were cited as potentially useful means of support in addition to counselling and better awareness of NF1 amongst the general population. Information about NF1 had most commonly been sought from the internet (67%, n=49) followed by medical staff (49%, n=36) and then support groups (47%, n=34).

Four areas were mentioned both as avenues of support and information. Friends were an important source of support but not information. More participants cited family as a source of support than as a site of information. The greatest information source was the internet and support groups were used by people as an information source rather than as a source of support.

<b>Source</b>	<b>Information</b>	<b>Support</b>
Friends	2.7 %	58.9%
Family	26%	64.4%
Internet	67.1%	23.3%
Support group (The Neuro Foundation/helpline/other groups)	50.7%	8.2%

Table 9: Sources of information and support reported by young people

Participants were fairly evenly divided between agreeing (52.3%, n=34) and disagreeing (47.7%, n=31) that they had or have good support at school. However, most (79.1%, n=49) agreed that their school did not really understand NF1. Twenty five participants provided illustrative comments on

school experience. These were analysed and four codes were revealed. Appearance was cited as important within social interactions for some participants. Codes and illustrative quotes are provided in table 10.

Code	% and number of responses	Example of quotes from open question
Learning difficulties and support in school	28% (n=7)	<i>'I struggled with some of the subjects, found it hard to keep up with my friends doing the work' (respondent 14)</i>  <i>'it was a long hard battle for my parent to get the necessary support at school'(respondent 44)</i>
Negative social interactions	16% (n=4)	<i>'I had very hurtful pictures and comments said and spread about me at school' (respondent 7)</i>  <i>'got teased about a huge tumour on my face constantly' (respondent 18)</i>
People not knowing what NF1 was	44% (n=11)	<i>'no one really knows what nf1 is unless you tell them (teachers and students)' (respondent 75)</i>
Positive experiences of school	12% (n=3)	<i>'I had lots of support in high school they understood when I have had time off school for appts and surgery' (respondent 76)</i>  <i>'I've always excelled in school. I guess in that respect, I am lucky. Working towards earning a master's degree at the moment.'(respondent 33)</i>

Table 10: Comments about school: a content analysis of young people's survey responses

When asked which professionals they had seen in relation to NF1, participants identified 12 types of medical professions (GP, geneticist, eye specialist, NF1 specialist, paediatrician, dermatologist, neurologists, pain specialists, ENT, orthopaedics, plastic surgeon and neuro surgeon). Three (4.1%) did not know who they saw and six (8.2%) reported that they did not see anyone. Participants were evenly split between agreeing (50%) and disagreeing (50%) that health professionals generally know about and understand NF1. Most (80.5%, n= 58) sometimes had to explain what NF1 is.

Twenty eight respondents added further comments regarding health professionals. Analysis revealed three distinct codes, the most common related to a perceived lack of knowledge and understanding of NF1 by some health professionals, although comments also indicated that this varied depending on whether or not specialists were seen. Codes and illustrative quotes are provided in table 11.

Code	% and number of responses	Example of quotes from open question
Positive comments re health professionals	25% (n=7)	<p><i>'The doctors that have cared for me have been extremely knowledgeable and have told me about different studies that are underway.'</i> (respondent 22)</p> <p><i>'my nurse is really understanding she helps me fully understand anything they are talking about'</i> (respondent 57)</p>
Negative comments re health professionals	32% (n=9)	<p><i>'The doctors and nurses I had made thing worse for me they made me feel like I was some kind of lab rat and when they talk it was to my parents'</i> (respondent 64)</p> <p><i>'.....they treat me like a freak show and I have the trainee drs in and out of my room without permission'</i> (respondent 7)</p>
Professionals not knowing about or understanding NF1	43% (n=12)	<p><i>'A lot of the time I say to Drs or nurses I have NF, and they look very confused and say what's that?'</i> (respondent 11)</p> <p><i>'Health visitors and midwives have no idea about it! When I was pregnant with my 2 Yr old, I had to keep explaining myself to them..... Not helpful when I'm worried that our unborn baby could have it'</i> (respondent 48)</p>

Table 11: Comments about health professionals: A content analysis of young people's survey responses

### Summary of young people's survey findings

Analysis of the interviews with young people indicated that young people's wellbeing (happiness) was associated with their feelings about their appearance in general (but not the noticeability of their NF1), their experience of social situations and stigma, and their perceptions of NF1 (see figure 5 in Chapter 4). These findings informed the choice of research questions and specific measures used in a survey to explore these issues in a larger sample and to examine the extent to which illness threat (B-IPQ), stigma (PSQ), social comfort (SCQ) and body esteem (BE App) predict happiness.

Happiness was, as proposed, related to general feelings about appearance, young people's social comfort and stigma experience and their perceptions of NF1. The best predictor of young people's happiness was general feelings about appearance. Aspects explored in the survey were grounded in earlier interview data, in order to ensure these areas are representative of young people's experience. However, it is important to note that there may be other aspects of experience that impact on wellbeing, for instance analysis of young people's concerns in the survey highlighted future family plans as being a concern that was not prevalent during many interviews. This may be an issue that concerns young people, however it was not discussed by the interviewer during interviews due to ethical considerations (See section 3.4). Therefore young people may have not had an opportunity to voice this as a particularly important aspect of their experience. It may also be that young people feel more comfortable highlighting this issue in a survey rather than in a one to one interview.

Findings presented in this chapter support the earlier observation that the wellbeing of young people with NF1 was not related to their perception of whether or not their condition was noticeable. Whether young people reported their condition as noticeable or not was not found to be relevant within their perceptions of stigma, social comfort, illness perceptions, body esteem or happiness.

While no significant differences were found in overall appearance scores for those who considered their NF1 noticeable or not, the way young people felt about their appearance overall was significant to their experience and happiness. Body esteem (appearance subscale) scores correlated highly with all other measures and a regression analysis identified appearance as a highly significant predictor of young people's happiness. This finding suggests that it is satisfaction with general appearance that is important to young people with NF1 rather than the subjective noticeability of their condition.

Young people reported concerns regarding other people's reactions to the visible signs of their condition; they described covering visible marks and recounted difficulties related to others not having heard of NF1. The lowest scores in the social experience section of the PSQ related to the absence of friendly behaviour rather than staring or hostility, suggesting that rather than overt hostility, young people with NF1 sense others around them being generally unfriendly and unsure of how to respond to a visible difference.

Findings highlight the individual and variable nature of NF1 and the many different ways that young people experience the condition. Some described NF1 as a 'living hell', a 'curse' and a 'misery' while others wrote that it was 'just part of my life' and 'part of who I am and I wouldn't change it for anything'. These comments and the range of scores on the measures used in the survey reflect the individual and subjective nature of living with NF1.

### 8.5 Parents' survey findings

Fifty five parents completed the questionnaire, (32 responses online, 23 on paper). The majority of parents were female (85.5%, n=47) and 94.5% (n=52) were White British, American or Irish, one person was of mixed ethnicity, one was Black African Caribbean/Black British/American and one preferred not to answer. Over half of parents (58.2%, n=32) lived in England, 25.5% (n=14) lived in the USA. The remaining parents lived in Scotland (3.6%, n=2), Wales (3.6%, n=2), Canada (5.5%, n=3), New Zealand (1.8%, n=1) and Mexico (1.8%, n=1). All participating parents had a child aged 14-24 with NF1. 45.6% (n=24) of these children were male (1 person did not provide this information). Just over half (56.3%, n=31) of participants had children aged under 18.

Twenty three parents (41.8%) had a diagnosis of NF1 themselves. 43.6% (n=24) reported that their child's NF1 was inherited and 52.7% (n=29) said it was new to the family whilst two people were unsure. The majority of parents (70.9%, n=39) reported that their child was diagnosed before the age of five. Parents reported their child having been seen by a variety of medical professionals, most frequently GP's/family doctors (43%), geneticists (43%) and eye specialists (41.4%). A total of 22 different types of professionals were listed by parents; one-third reported their child had seen an NF1 specialist.

The table below provides descriptive statistics for the four standardised measures (including subscales where appropriate) used in this survey. Findings are discussed in the following sections in relation to the research questions outlined in section 7.2.

Scale	N	Min	Max	Mean	Possible Range	Std. Deviation
Brief-Illness Perceptions Questionnaire-Parent on child) (higher score indicates more threatening view of NF1)	48	24	74	52.40	0-80	11.749
Brief-Illness Perceptions Questionnaire-parent (higher score indicates more threatening view of NF1)	50	22	78	51.34	0-80	11.559

PSQ subscale1 (absence of friendly behaviour) (Higher scores indicates higher levels of perceived stigma)	47	1.00	4.50	2.3404	1-5	.72966
PSQ subscale 2 (Confused/staring behaviour) (Higher scores indicates higher levels of perceived stigma)	47	1.00	4.00	1.7660	1-5	.82200
PSQ subscale 3 (Hostile Behaviour) (Higher scores indicates higher levels of perceived stigma)	46	1.40	4.20	2.4087	1-5	.71300
Perceived Stigma Questionnaire (Higher scores indicates higher levels of perceived stigma)	43	1.00	4.00	2.0875	1-5	.71481
Social Comfort Questionnaire (higher scores indicate higher levels of social comfort)	48	2	5	3.11	1-5	.848
Coping Health Inventory for Parents FAMILY (higher scores indicates greater helpfulness of the coping pattern)	46	8	46	23.26	0-48	9.434
Coping Health Inventory for Parents SOCIAL (higher scores indicates greater helpfulness of the coping pattern)	41	0	48	18.22	0-54	8.943
Coping Health Inventory for Parents UNDERSTAND (higher scores indicates greater helpfulness of the coping pattern)	46	4	24	10.59	0-24	4.874

Table 12: Descriptive scores on the standardised measures used in the survey of parents

### How do parents describe the role of appearance within their child's experience of NF1 and how noticeable do they feel their child's condition is?

The majority of parents (79%, n=42) reported their child rarely/never expressed concern regarding their appearance in general and 85% (n=45) said their child rarely/never expressed concern about appearance-related aspects of NF1. Most parents (66%, n=24) were fairly confident (6-10 on a scale of 0-10, with 10 being very confident) in managing their child's appearance concerns. However 34% (n=12) of parents indicated medium to low levels of confidence (0-5 on the scale). The majority of parents (60%, n=32) felt their child's attitude towards their appearance had not really changed at any point. Of those who did feel their child's attitude had changed, many thought this was due to them being a teenager (46% n=13), so concerns about appearance were normalised and not NF1 specific.

Parents were asked to briefly describe the main ways NF1 affected their child, 55 parents responded. Using content analysis answers were coded into categories, as shown in table 13

<b>Code</b>	<b>% and number of responses</b>	<b>Example of quotes from open question</b>
Educational	25.5 % (n=14)	'She has learning problems due to the condition. She does find school quite challenging at times' (respondent 2).
Medical	23.6 % (n=13)	'tumors on spine' (respondent 47).
social concerns	18.2% (n=10)	'Difficulty making friends and maintaining friendships' (respondent 37)
Appearance	18.2% (n=10)	'She doesn't think she beautiful, hates the cafe au lait spots and lumps' (respondent 13)
employment and careers	1.8% (n=1)	'couldn't do they job they want' (respondent 29)
the uncertainty of the condition	7.3% (n=4)	'uncertain future, increased stress' (respondent 58)
reported that NF1 had no particular effect on their child.	5.5% (n=3)	'no real problems' (respondent 10)

Table 13: Main way NF1 affects child: A content analysis of parent's survey responses

Parents were asked about the effect of their child's NF1 on themselves. Content analysis showed the greatest effect to be a general sense of worry, codes and illustrative quotes are provided in table 14.

<b>Code</b>	<b>% and number of responses</b>	<b>Example of quotes from open question</b>
general sense of worry and monitoring their child's symptoms	41% (n=21)	"....worry.....he is now approaching 14 so very aware that lumps and bumps grow during puberty (respondent 11)."  "Always keep in the back of my mind" (respondent 17).
managing learning and behavioural difficulties	26% (n=13)	"The learning disabilities and behavioural problems effect our daily lives" (respondent 37).
the impact on career and managing medical appointments	10% (n=5)	'I cannot work as my daughter has a lot of time off school, it is stressful and challenging" (respondent 5)

		<i>'never ending hospital appointments'</i> (respondent 54).
Guilt	8% (n=4)	'it breaks my heart that I passed this onto my children' (Respondent 15)
Specific medical concern	6% (n=3)	'Scoliosis' (respondent 55)
No Impact	10% (n=5)	".....we continue as if we did not have the diagnosis" (respondent 27).

Table 14: Main way NF1 affects self: A content analysis of parent's survey responses

The majority of parents (n=35, 59%) reported their initial concerns at the time of their child's diagnosis primarily related to understanding the condition and the medical prognosis *'How is this going to affect her'* (respondent 3) and *'Was he going to die? What kind of life was he going to have?'* (respondent 19). In contrast parents were now mostly concerned about their child being generally happy and living a normal adult life (n=26, 43%) as illustrated by the following; *"Will she ever have a friend to enjoy life with? Will she ever leave home? Will she be able to work and feel a sense of achievement and self worth?"* (respondent 2) and *"That he has as normal a life as possible"* (respondent 21) and *"How can I help her to have the best future possible?"* (respondent 37).

Parents were asked to rate the noticeability of their child's NF1. Over one quarter of parents (28%, n=15) felt their child's NF1 was not at all noticeable to others (scoring 0). The exact same number of people felt their child's NF1 was noticeable over the midpoint (between 6-10). The mean noticeability score was 3.57 (SD 3.220).

#### **How do parents report their child's social comfort and interactions with others and does this relate to parents' reports of noticeability?**

Parents' perceptions of their child's social comfort and perceptions of stigma were measured, as in the young people's survey, using the PSQ and SCQ. Scores relate to the parent's view of their child's experience. Mean PSQ scores (table 12) indicate parents perceive their children to experience low to moderate levels of stigma in all subscales. Total PSQ scores indicate most parents (n=30, 70%) never or almost never perceive stigma while 9% (n=4) perceive that their child often experiences stigma. The greatest levels of reported stigma related to hostile behaviour, 43.5% (n=20) of parents reported their child faced this type of stigma sometimes or often, one person (2.2%) reported this never occurred and 54.3% (n=25) that it almost never occurred. Absence of friendly behaviour was reported as occurring sometimes, often or always by 38.2% (n=18) of parents (53%, n=25) reported almost never and 8.5%, n=4 never). Finally, 23.4% (n=11) of parents reported that their child faced confused and staring behaviour, 27.7% (n=13) report this happened almost never and 48.9%, (n=23) reported that it never happened.

Scores on the SCQ indicate parents are fairly evenly divided between feeling their children almost never feel socially comfortable (25%, n=12), sometimes feel comfortable (37.5%, n=18) and often feel comfortable (31.3%, n=15), with no parents reporting never and 6.3% (n=3) reporting always.

The relationship between noticeability (measured on a scale of 0-10 with 0 being not at all noticeable) and the PSQ (and subscales) and SCQ was investigated using Pearson product moment correlation coefficient. Preliminary analysis was performed to ensure no violation of the assumptions of normality, linearity and homoscedasticity (descriptive data is reported in table 12). There was a medium positive correlation between noticeability and PSQ subscale 1 (absence of friendly behaviour) and a strong positive correlation between noticeability and PSQ subscales 2 (confused/staring behaviour) and 3 (hostile behaviour) and the PSQ total score. High noticeability was positively associated with higher levels of perceived stigma. There was also a strong negative correlation between noticeability and the SCQ total score. High noticeability was negatively associated with social comfort ie low levels of social comfort.

	R
PSQ 1: Absence of friendly behaviour	.347
PSQ 2: Confused/staring behaviour	.757**
PSQ 3: Hostile behaviour	.682**
TOTAL PSQ	.729**
TOTAL SCQ	-.590**

\*\* . Correlation is significant at the 0.01 level (2-tailed).

Table 15: Survey of parents: Correlations between noticeability and the PSQ and SCQ

How do parents report that they cope with their child having NF1? Are there specific ways in which parents cope and do coping patterns relate to whether or not the condition was inherited?

This section relates to parents' reports of coping with and managing their child's condition, using the Coping Health Inventory for Parents (CHIP) and asking about issues identified in the interviews as impacting on general day-to-day management of NF1 (including parents' interactions with health professionals, school, information and support needs and media portrayal of the condition).

The CHIP is comprised of three patterns: pattern one focuses on behaviours that strengthen family life and relationships and parents outlook on life/optimism, pattern two focuses on parents' efforts to develop relationships with others and engage in activities that enhance their individual identity and self worth as well as behaviours that manage tension and pressure, pattern three concentrates on parents' relationships with healthcare professionals and other parents of chronically ill children. Respondents are asked if a series of coping behaviours are not helpful (0), minimally helpful (1),

moderately helpful (2) or extremely helpful (3), or respondents can indicate a behaviour was 'not possible' or 'chose not to use it'. Descriptive statistics for the CHIP subscales are provided in table 11.

The highest scoring coping behaviours (as a percentage of total score, see table 16) were "reading more about NF1" and "doing things with my children" while the lowest scoring behaviour was "taking care of medical equipment at home" (table 16) although this may not be pertinent to parents of young people with NF1 as the condition does not often necessitate medical equipment at home.

Behaviour	Total score	Total possible score	Percentage of total score
<b>&gt;score of 60 and 40%</b>			
Reading more about NF1	65	159	41%
Doing things with my children	61	150	41%
Having my child with NF1 seen at the hospital/GPs on a regular basis	64	159	40%
Talking with medical staff	63	156	40%
<b>&lt;score of 30 and 20%</b>			
Getting other members of the family to help with chores and tasks at home	27	153	18%
Eating	20	153	13%
Buying gifts for myself and/or other family members	13	153	8%
Taking care of medical equipment at home	8	147	5%

Table 16: Survey of Parents: Highest and lowest scoring coping behaviours reported on the CHIP

In order to investigate differences in the way parents utilised coping patterns the mean score of each scale was calculated by dividing the score on the scale by total possible score on the scale (table 16). Calculating the mean in this way with the CHIP scale has been used previously in similar research with parents of children with Juvenile Idiopathic Arthritis (JIA) (Cavallo et al, 2009). Coping pattern three (parents' relationships with healthcare professionals and other parents of chronically ill children and their knowledge and understanding of NF1) received the highest percentage of total possible score. The second highest scoring pattern was coping pattern one, behaviours that strengthen family life and relationships as well as parents' outlook on life/optimism. The lowest scoring coping pattern was two, this focuses on parents' efforts to develop relationships with others and engage in activities that enhance their individual identity and self worth as well as behaviours that manage tension and pressure.

Scale	Mean score	SD	Total possible score	% of total possible score (mean/total possible)
CHIP1 FAMILY	23.26	9.434	57	40.81
CHIP2 SOCIAL	18.22	8.943	54	33.74
CHIP3 UNDERSTAND	10.59	4.874	24	44.13

Table 17. Survey of parents: Mean CHIP scores and % of total score

The CHIP was included in the questionnaire partly to investigate if coping patterns are different between families where a diagnosis of NF1 is new (i.e. a spontaneous mutation) or inherited from a parent. In order to examine this, scales were examined as above investigating percentage of total possible scores for families where the condition was new or inherited. As can be seen in table 18, below, regardless of whether diagnosis is new or inherited the scales follow a similar pattern to the overall total possible score as reported above. However coping behaviours on subscales one and three are reported as less helpful by parents where the condition is inherited.

Scale	% of total possible score	% of total possible score <b>New</b>	% of total possible score <b>Inherited</b>
CHIP1FAMILY	40.81	44.67	37.19
CHIP2SOCIAL	33.74	33.94	34.15
CHIP3UNDERSTAND	44.13	48.5	42.04

Table 18: Survey of parents: mean CHIP scores and % of total score reported by families where diagnosis was new to the family or inherited

### **What are parents' perceptions of NF1 and do these relate to the perceived noticeability of their child's condition?**

The B-IPQ was firstly used to assess parents' direct perceptions of NF1. As in the young people's survey the mean timeline score on the B-IPQ was high (9.83, SD .706), this high score may again be indicative of the genetic nature of NF1; as in the young people's survey, several participants commented that this question was pointless. A mean score of 2.17 (SD 2.75) on the control item suggests minimal personal control over their child's condition; 44.4% (n=24) of parents reported a score of 0 (absolutely no control) on this item. Although still minimal, the mean score for treatment control was slightly higher (m 3.89 SD 2.72) and fewer parents (18.9%, n=10) scored 0. The mean identity score was 5.44 (SD 2.96), and 7 parents (13%) scored 10 (many severe symptoms). Concern had a mean score of 7.40 (SD 2.85) and 21 (39.6%) scored 10 (extremely concerned). In terms of coherence, the mean score for parents' own understanding was 7.37 (SD 2.41) suggesting a good understanding of NF1. Emotional representation had a mean score of 6.15 (SD 3.17). However, 20% (n=11) of parents indicated they were extremely affected emotionally (scoring 10). The causal section of the Brief IPQ asked parents to list the three most important things that they believed caused their child's NF1. 87.2% of parents who completed this section wrote that the cause was genetic.

Complications in pregnancy, treatment, parental age, incest and an accident were also cited. Three parents were unsure of the cause.

The B-IPQ in this survey also included additional questions so that illness perceptions could also be investigated in terms of parents' views of their child's illness perceptions. Three questions were altered to ask about 'your child' as well as 'you'. B-IPQ question one asked parents about the effect of NF1 on their child's life, the mean score of 5.87 (SD 2.997) indicates a moderate affect, however within this 50.1% of parents scored 7 or above indicating NF1 severely affects their child, 27.9% scored 0-3, indicating low or no affect and 22.3% scored in the midrange between 4 and 6. B-IPQ question seven asked parents about their child's understanding of NF1, the mean score of 5.63 (SD 3.018) indicates a medium level of understanding however within this 39.9% of parents scored 7-10, indicating their child has a good level of understanding. B-IPQ question eight asked parents about the emotional effect of NF1 on their child. The mean score was 5.21 (SD 3.304), indicating a medium affect. Again scores varied greatly from 0 to 10.

A paired samples t test was conducted to evaluate the difference between overall scores on the B-IPQ, relating to parent's perception of (a) impact on parent and (b) perceived impact on child. No significant difference was found between impact on parent (m 51.34 SD 11.559) and perceived impact on child (m 52.40 SD 11.749). A paired sample t test was also conducted to evaluate the difference between responses on the three items that had been altered (questions 1,7 & 8 as indicated above). No significant difference was found on the consequence item between the parent scores 'How much does NF1 affect your life?' (m 6 SD 3.003) and perceived impact on child 'How much does NF1 affect your child's life?' (m 5.87 SD 2.997). There was a statistically significant difference in scores on the coherence item between the impact on parent 'How well do you feel you understand NF1?' (m 7.37 SD 2.413) and perceived impact on child 'How well do you feel your child understands NF1?' (m 5.63 SD 3.018),  $t(53) = 3.872$ ,  $p < .0005$  indicating that parents thought they understood it better than their child. The impact on self/parent was reported as greater than the impact on child. There was also a statistically significant difference in scores on the emotional item between the impact on parent (m 6.15 SD 3.165) and perceived impact on child (m 5.21 SD 3.304),  $t(51) = 2.473$ ,  $p < .05$ . The impact on self/parent was reported as greater than the impact on their child.

The relationship between noticeability and the B-IPQ was investigated using Pearson product moment correlation coefficient. There was a medium positive correlation between noticeability and the B-IPQ. High noticeability was positively associated with high threat levels for parents themselves and their child (Brief IPQ).

	R
B- IPQ (Parent)	.425**
B- IPQ (Parent on Child)	.491**

\*\* . Correlation is significant at the 0.01 level (2-tailed).

Table 19: Survey of parents: Correlations between noticeability and the B-IPQ

### How do parents' perceptions of NF1 relate to their views of their child's social comfort and interactions with others?

The relationship between the B-IPQ and the PSQ and SCQ was investigated using Pearson product moment correlation coefficient. Preliminary analysis was performed to ensure no violation of the assumptions of normality, linearity and homoscedasticity. Correlations between the B-IPQ, PSQ and SCQ are detailed in table 20 below. The high correlation between B-IPQ C and B-IPQ P (C=parents' perceptions of impact on child, P=parents' perceptions of impact on self) would be explained by the commonality of questions in these measures (only three questions differed as described in section 7.3.2.4). Higher IPQ scores indicate a greater perceived threat for parent/child. Both IPQ scores correlated positively with perceived stigma and negatively with social comfort, suggesting a relationship between higher perceived threat on the brief IPQ and higher perceived stigma on the PSQ and lower social comfort on the SCQ.

Scale	1 B-IPQ p	2 B-IPQc	3 PSQ	4 SCQ
1 B- IPQ p	-	.906**	.357*	-.356*
2 B-IPQ c		-	.432**	-.400**
3 PSQ			-	-.800**
4 SCQ				-

\*\* . Correlation is significant at the 0.01 level (2-tailed).

\* . Correlation is significant at the 0.05 level (2-tailed).

Table 20: Survey of parents: Correlations between scores on B-IPQ, PSQ and SCQ

### How do parents describe their (and their child's) information and support needs, and how is appearance mentioned within this?

Parents were asked if they agreed or disagreed with a series of quotes from the interviews about their child's school, medical staff, representations of NF1 in the media, and awareness of NF1 (see table 21). Most parents indicated that their child did not have good support at school and that school did not understand the condition. Over two thirds agreed that getting the right support was a 'battle' and the majority felt that NF1 did make a difference to their child at school. Most felt medical staff did not know about NF1 and had to sometimes explain the condition and be the expert. The vast majority felt that most people have not heard of NF1 although they were happy to explain it and they agreed that having the condition would be easier if others knew about it. While the majority felt the internet was a good place to find information about NF1, most also felt TV, magazines and the internet did not show realistic examples of the condition, although most reported that charities and support groups do show positive images of people with NF1.

	<b>Agree</b> N (%)	<b>Disagree</b> N (%)
My child had/has good support at school	14 (40%)	21 (60%)
My child's school didn't/doesn't understand NF1	29 (88%)	4 (12%)
It was a battle to get the right support for my child at school	23 (68%)	11 (32%)
NF1 didn't make any difference to my child at school	10 (29%)	25 (71%)
Medical staff generally know about and understand NF1	11 (31%)	24 (69%)
I sometimes have to explain to medical staff what NF1 is	27 (75%)	9 (25%)
Most medical staff don't know about NF1, I have to be the expert	26 (72%)	10 (28%)
Most people have normally heard of NF1	3 (9%)	32 (91%)
I'm happy to explain NF1 to other people	34 (94%)	2 (6%)
TV magazines and the internet show realistic examples of NF1	5 (14%)	31 (86%)
The internet is a good place to find information on NF1	27 (75%)	9 (25%)
Having NF1 would be easier if more people knew about it and understood it	31 (86%)	5 (14%)
Charities & support groups show positive images of people with NF1	27 (75%)	9 (25%)

Table 21: Survey of parents: Parents' reports of experience of managing NF1 with regards to school, general understanding and representations of the condition and interactions with medical staff

Parents reported seeking information and support from various sources, 78.2% (n=43) had used the internet, 72.7% (n=40) cited The Neuro Foundation, 52.7% (n=29) medical staff and 47.3% (n=26) specifically sought information and support from a NF specialist advisor. Online support groups were used by 29.1% (n=16) of parents. Three parents had never searched for information or support.

Parents were asked what support, if any, they would like to see available for (a) parents and (b) young people with NF1 (table 22). The most frequent response was targeted information and support for parents (32.7% n=18) and young people (41.8% n=23). They particularly wanted to meet other parents in a similar position and for their children to have the chance to meet others of the same age.

	Support for parents		Support for young people	
	Frequency	Percent	Frequency	Percent
information and targeted support	18	32.7	23	41.8
easy access to healthcare and specialists	6	10.9	3	5.5
someone (professional) to talk to	4	7.3	7	12.7
general better awareness	4	7.3	3	5.5
better educational support	7	12.7	2	3.6
Financial	2	3.6	n/a	n/a
Social Skills support	n/a	n/a	2	3.6

Table 22: Survey of parents: Parents accounts of the types of support desired for themselves and young people

### **Summary of findings from the parents' survey**

The interview findings in Chapter 5 suggested that parents' perceptions of the overall impact of NF1 both on themselves and their child were associated with their child's management and experience of social situations and perceptions of NF1 and the appearance and noticeability of their child's condition.

In this survey, perceptions of NF1 were found to be associated with parents' assessments of their child's management and experience of social situations and social comfort. Parents reported a range of coping patterns which did not differ for families where NF1 was new or inherited. Additionally, as proposed, greater perceived noticeability was associated with greater perceived impact of NF1 for the parent and child.

Parents' responses indicate that NF1 impacts on and affects their own life and their child's. Parents believed that they understood NF1 better than their child did and that the condition had a greater emotional impact on them than their child. Their qualitative responses indicated that they viewed the impact of NF1 on their child as primarily educational and medical, although over a third felt the greatest impact was social and appearance-related.

Parents seemed to normalise their child's appearance concerns, and most described these as part and parcel of being a teenager. However, while some parents reported that their child rarely or never expressed concern about their appearance (either in general or in relation to NF1), the noticeability of their child's condition was important to parents. Noticeability was associated with parental reports of illness threat (to self and child), perceived stigma and social comfort.

Parents reported their child's NF1 as being an on-going concern; it was always in the back of their mind and a permanent yet fluctuating source of anxiety. It may be this sense of anxiety or worry that makes the noticeability of NF1 important within parents' experiences; watching for the emergence of new physical signs of NF1 may be one way in which parents monitor the progression of their child's condition which in turn heightens their awareness of the noticeability of the condition.

Parents described a pattern in which their early concerns (at initial diagnosis) were related to understanding NF1 and worrying about the medical consequences whilst, over time, concerns were more related to wanting their child to live a 'normal life'. Whilst coping patterns varied greatly between individuals, most involved relationships with health professionals and other parents of chronically ill children, and gaining an understanding of the condition.

## 8.6 Comparison of parents' and young people's responses

### How do parents and young people differ on their descriptions of social comfort, interactions with others and perceptions of NF1?

This section presents findings related to whether or not young people and parents report similar illness perceptions, stigma and social experience as measured on the B-IPQ, PSQ and SCQ.

An independent samples t test was conducted to investigate the relationship between parents' B-IPQ scores and young people's B-IPQ scores. No significant difference was found between scores for parents reporting on their child ( $m=52.4$ ,  $SD 11.7$ ) and young people themselves ( $m= 48.3$ ,  $SD=11.5$ ;  $t(115) =1.87$   $p= 0.64$  two tailed) The magnitude of the differences in the means (mean difference =  $4.09$ ,  $95\% CI: -0.235$  to  $-8.418$ ) was small.

An independent samples t test was conducted to investigate the difference between parental and young people's reports of young people's stigma experience and social comfort. No significant difference was found between scores on the PSQ for parents ( $m=2.2$ ,  $SD 0.585$ ) and young people ( $m= 2.09$ ,  $SD=0.715$ ;  $t(99) =0.81$ ,  $p= 0.421$  two tailed) The magnitude of the differences in the means (mean difference =  $-0.105$ ,  $95\% CI: -0.362$  to  $0.152$ ) was small. Furthermore no significant difference was found between scores on the SCQ for parents ( $m=3.1$ ,  $SD 0.848$ ) and young people ( $m=3.1$ ,  $SD=0.778$ ;  $t(111) =0.07$   $p= 0.944$  two tailed) The magnitude of the differences in the means (mean difference =  $0.011$ ,  $95\% CI: -0.294$  to  $0.315$ ) was small.

## 8.7 Health Professionals' survey findings

In total 53 surveys were completed by health professionals (49 online). Twenty nine respondents (55%) worked in the UK, four (7.5%) in Australia, four (7.5%) in the USA, five (9.4%) in Canada and one (1.9%) in The Republic of Ireland. Ten (18.9%) participants did not provide this information. Forty five percent ( $n=23$ ) of respondents were genetic counsellors, 21.6% ( $n=11$ ) were geneticists, the remaining 33.3% ( $n=17$ ) included dermatologists, nurses, nurse specialists, paediatricians, endocrinologists, researchers, and psychologists. Four participants (8%) reported seeing a patient with NF1 once a year or less, 29 (56%) saw a patient with NF1 several times a year, while 19 (36%) saw patients monthly, weekly and daily.

### How do health professional describe NF1 impacting on young people and how is the role of appearance described within this?

Most health professionals (82%,  $n=43$ ) agreed that the way people manage their condition depends on who else in the family has NF1 and 84% ( $n=45$ ) believed parents' concerns depended on who else in the family was affected. Health professionals were asked to describe the most common concerns

for families where a diagnosis of NF1 was new to the family or when other members already had the condition. When NF1 was new to a family respondents reported understanding the medical prognosis (38% n=18) as the greatest concern followed by the child's appearance (30% n=14) and uncertainty (11% n=5). Other concerns identified were inheritance (specifically who else may have the condition, 9% n=4), medical management (6% n=3), wanting information (4% n=2), and how to explain NF1 to their child (2% n=1).

When NF1 was a pre existing condition in a family respondents reported that concerns were the severity of NF1 (including in relation to other affected family members, 34% n=14) while 24% (n=10) responded that concerns were highly dependent on the family's experience of NF1. Twelve percent (n=5) cited appearance as the main concern, 10% (n=4) cited medical management, inheritance and genetic testing (7% n=3) and long term medical prognosis (5%, n=2). The following were each cited by 1 health professional (2%); education, guilt and quality of life.

Health professionals reported the most common concern for young people with NF1 was its impact on their appearance (61%, n=25). The second most common concern was thought to be the future and uncertainty (17%, n=7) while the remaining concerns related to employment (2% n=1), having children (2% n=1), social concerns (10%, n=4), health insurance (2% n=1), and wanting to get on with life (5% n=2).

Health professionals were asked if they agreed or disagreed with a series of quotes from earlier interviews about NF1 and appearance (see table 23). Findings suggest that health professionals perceive appearance to be an important concern for young people with NF1. The majority agreed that *'Adolescence is a time people start to worry about appearance'* and that *'most young people with NF1 are worried about how they look'*. About half of those surveyed reported that appearance evaluations change for young people with NF1 during adolescence. Uncertainty was highlighted as an important concern and most health professionals (62%, n=33) agreed that *'Tumour load is not an indicator of how an individual with NF1 feels about their appearance'*.

<b>Statement</b>	<b>% (n) Disagree or strongly disagree</b>	<b>% (n) Neither agree or disagree</b>	<b>% (n) Strongly agree or agree</b>
Young children with NF1 are normally positive about their appearance	7.5 (4)	45.3 (24)	47.2 (25)
Adolescence is a time people start to worry about appearance	1.9 (1)	5.7 (3)	92.4 (49)
Most young people with NF1 are worried about how they look	1.9 (1)	24.5 (13)	73.6 (39)
People with NF1 change how they view their	7.5 (4)	43.4 (23)	49 (26)

appearance between childhood and adulthood			
It's hard for young people with NF1 to talk about appearance	18.9 (10)	37.7 (20)	43.4 (23)
Young people with NF1 worry about neurofibromas appearing	5.7 (3)	18.9 (10)	75.5 (40)
The uncertainty of NF1 is the biggest concern for many young people	7.7 (4)	26.9 (14)	65.4 (34)
Tumor load is not an indicator of how an individual with NF1 feels about their appearance	11.3 (6)	26.4 (14)	62.3 (33)
Parents are often concerned about how to prepare children for possible changes to appearance	3.8 (2)	18.9 (10)	77.4 (41)
It is difficult for all young people to talk about their appearance	30.8 (16)	19.2 (10)	50 (26)

Table 23: Survey of health professionals: Reports of NF1, appearance and adolescence

When asked why they thought concerns about appearance changed during adolescence, health professionals agreed with a range of reasons that had been identified in interviews such as the presence of neurofibromas (77%, n=41), dating (74%, n=39), friends (71%, n=38), the media (63%, n=33) and celebrity culture (51%, n=27). Participants also mentioned that appearance concerns could relate to hormones, use of facebook, individual confidence, uncertainty, family and individual differences. Additionally 88% (n=47) of health professionals thought appearance concerns were a normal part of being a young person rather than being NF1 specific.

All of the aspects considered in this section were explored to investigate whether there were differences in health professionals answers based on their professional role or nationality. There were no clear differences in how different groups described the impact of NF1 on young people and the impact of appearance within this. However it is important to note that it is difficult to come to a clear conclusion regarding this as 19% of respondents did not provide their nationality and the way in which roles were described was not always clear.

### **What psychosocial support do health professionals feel would help young people with NF1 to adapt positively to NF1 and the appearance related aspects of the condition?**

Health professionals were asked about the delivery of support for young people with NF1. Online support, information for young people and supporting families early on are highlighted as needed (Table 24).

Type of support	Yes	Maybe	No
Local groups for young people	52%	48%	0
Online	84.6%	15.4%	0
Transition groups	51%	43.1%	5.9%

Transition programme	57.1%	36.7%	6.1%
Information	92.3%	7.7%	0
Camps	46.2%	44.2%	9.6%
Helplines	59.6%	34.6%	5.8%
Support families early	70.6%	29.4%	0

Table 24: Survey of health professionals: Types of support needed for young people

Accounts of ease of access to support services (for young people) varied considerably. Just over half of health professionals (58%, n=30) stated that their service had links with a psychological service and 45.3% (n=34) felt there was a clear pathway for psychological support. However 46%, (n=24) thought it was hard to access these services. Over a third (n=19) of participants thought that patients were involved with support groups. Just over half considered the internet useful for finding information on NF1 (58.5%, n=30).

### **How do health professionals describe their role in young people's management of NF1?**

Health professionals were asked about their contact with young people with NF1 and whether this changed during adolescence. Fifty two percent (n=26) of respondents tended to see people with NF1 at a younger age or as adults. In terms of engagement in healthcare, 21% (n=10) agreed that 'Very few young people will actively manage their healthcare' and 29% (n=14) reported it was difficult to reach young people. The majority of health professionals (75.4%, n=40) agreed that an important part of their role was helping families to talk to their children about NF1 and 69.8% (n=37) agreed that parents are often concerned about how to do this. Just under half of participants (43.4% n=23) agreed that parents are best placed to give their children information about the condition and 64.2% (n=34) worried parents have not fully informed their children about NF1.

### **Summary of health professionals' survey findings**

In interviews, health professionals had reported that young people's experiences of NF1 were related to their appearance, psychosocial support and engagement with healthcare.

In the survey, health professionals reported appearance and the uncertainty of NF1 as the most common concerns for young people with NF1. Adolescence was seen as a time when concerns about appearance begin for all young people and respondents agreed that an objective assessment of appearance does not indicate how individuals feel about the way they look. Health professionals also reported that parents have concerns about preparing children for possible changes to their appearance and identified a range of possible reasons why appearance concerns increase during adolescence; it was part of growing up but also when changes such as neurofibromas started to grow.

Health professionals felt that psychosocial support for young people was needed but was not always easy to access. The support viewed as most beneficial was online support, information targeted at young people (and ensuring young people understood their condition), as well as helping families holistically from diagnosis onwards.

Young people's engagement with healthcare was highlighted in interviews as being important but difficult to achieve. This was also apparent in the survey; just 33% of health professionals agreed that young people would actively manage their healthcare.

Health professionals also agreed overwhelmingly that management and experience of NF1 varies depending on inheritance patterns and whether anyone else in the family has the condition. If NF1 was new to a family, then common concerns were related to prognosis and appearance whereas if NF1 was a pre existing condition concerns were related to severity and would depend on that family's experience of the condition. Thirty percent of health professionals identified appearance as the main concern where NF1 was new to the family, compared with 12% if it was a pre existing condition.

Findings from the survey of health professionals suggest that they perceive appearance to be an important (and complex) aspect within the management of NF1 during adolescence.

## **8.8 Conclusion**

The aim of the three surveys presented in this chapter was to explore the role of appearance within specific areas of young people's experiences of NF1 through their own reports, parents' reports and the reports of health professionals. The content of the surveys was determined by the earlier qualitative phase of this research.

Survey findings particularly highlight the variability in young people's experiences of NF1; for some, living with NF1 was extremely challenging, whilst others felt it had little impact on their lives. This was apparent in young people's, parents' and health professionals' responses. There was recognition across the surveys that management of NF1 is complicated and depends on family variables, social experience, understanding the condition and general feelings about appearance. All groups highlighted the importance of access to health professionals who know about and understand NF1.

It is important to note that the three groups differed in terms of how much of an impact appearance was thought to have. It was not young people's greatest concern and mean body esteem scores were not overly negative. Furthermore, subjectively reported noticeability was not important within young people's own reports. Yet body esteem scores were a strong predictor of young people's happiness. Parents reported that their children did not report appearance concern often, generally or in relation to NF1, yet their accounts of noticeability correlated with reports of illness perceptions, social comfort and perceived stigma. Health professionals reported that appearance was young people's greatest concern and considered appearance to be important, but few felt that tumor load was an indicator of appearance concern.

The following chapter provides a detailed discussion and evaluation of the key findings of both this and the previous qualitative stage and situates the findings of the PhD overall within the wider literature.

## **Chapter 9: Discussion**

This chapter considers the unique and original contribution this thesis makes to understanding the role of appearance within young people's experiences of NF1. The chapter synthesises, interprets and evaluates the findings from across this program of research, situating them within the wider literature in order to consider the role of appearance within young people's experiences of NF1. Following this, specific recommendations for the application of findings are considered, the methods used in this thesis are evaluated and finally suggestions for future research are outlined.

### **9.1 Research aim and summary of findings**

Adolescence and early adulthood is identified as an important developmental stage with regards to both health and appearance (eg Rumsey & Harcourt, 2007; Holmbeck, 2002). For young people with NF1 this may be a particularly challenging time due to the physical changes related to NF1 as well as social changes and a growing awareness of the appearance related aspects of the condition (Ferner et al, 2012; 2007). However the role of appearance within young people's experience has not previously been specifically examined in this population. This programme of research was undertaken in order to address this lack of evidence. The aim of the research was to provide an in depth exploration of the role that appearance plays within young people's day-to-day lived experience of Neurofibromatosis type 1 (NF1) from the perspective of young people themselves, their parents and health professionals working in this field. An exploratory mixed methods approach was chosen in order to investigate this previously unexplored area.

Findings presented in this thesis highlight the highly varied impact that NF1 may have on young people and the complex role that appearance plays in this. A relationship was found between young people's own appearance evaluations and their happiness, their perceptions of NF1 and their social interactions. However, their subjective accounts of whether or not their condition was noticeable to others were not found to be relevant within these aspects and appearance was not found to be young people's greatest concern. While some parents highlighted appearance as a key concern for their children with NF1, most reported that it was not. Parents' accounts of the noticeability of their child's NF1 did relate to their perceptions of NF1 and their child's social interactions. Health professionals emphasised the importance of appearance within some young people's experiences and reported that how individuals feel about their appearance does not necessarily relate to objective assessments. All three participant groups highlighted the social challenges associated with living with NF1.

### **9.2 Discussion of findings in relation to wider literature**

Across both stages of this research, reports of the importance and impact of appearance differed within and between participant groups. However within these varied accounts, the impact of appearance related to individual processes (such as appearance evaluations and managing

uncertainty) and social interactions (feeling comfortable socially and perceptions of stigma). These two key aspects will now be discussed in relation to the wider literature in the field.

### 9.2.1 Individual processes

Previous research with adults with NF1 has reported negative body image and appearance concerns (Smith et al, 2013; Granstrom et al, 2012). Furthermore there have been suggestions that young people with a chronic condition may have a less positive body image than healthy peers (Pinquart, 2013), although this may not always be the case (Lawrence, 2007). The mean body esteem scores for the young people in the survey were similar to means published by Mendelson et al (2001) in a normative population and by Lawrence et al's burns survivors and comparison group. This, in addition to just 15% of those surveyed in this research reporting negative body esteem, suggests that young people with NF1 do not have particularly low body esteem. A minority of parents reported that appearance was a concern for their child with NF1, and those who did highlight concerns often normalised them as part of being a young person and growing up. While health professionals reported appearance as young people's greatest concern, they also rationalised that there are pressures for all young people around appearance. Yet, while young people's body esteem scores were not particularly negative, possible future appearance changes were a concern and in interviews (in-line with previous suggestions from Ferner et al, 2007 & Ablon, 1999), they discussed a range of NF1 appearance-related concerns. Whilst at first these findings appear contradictory, on examination the appearance related concerns reported in interviews were also primarily related to the uncertainty of future changes, rather than young people's current appearance. The body esteem scores taken alongside the reported challenges young people discussed in the earlier interview stage suggest that whilst body esteem is important within young people's experience, it is not overly negative and appearance concerns relate to uncertainty regarding the condition and how their appearance could change. The unpredictability and uncertainty of NF1 means that appearance changes can happen throughout life. Managing this may therefore be a constant process regardless of actual change.

How individuals understand and feel about their appearance and condition is formed in many ways including interactions with health professionals, family experience, as well as socio-cultural factors. Illness perceptions are integral to how people manage an illness, Leventhal's self regulatory model (figure 11) proposes that perceptions impact on how individuals interpret, manage and cope with a health threat. Illness perceptions have not previously been explored with young people with NF1 and were used in the survey study in order to explore both young people's and parents' perceptions of NF1 in order to examine whether there was a coherent perception of NF1 and whether young people and parents held similar perceptions. Individual's illness perceptions influence their emotional response (Petrie & Weinman, 2006) and have been found to be an important aspect with the quality of life of individuals with other uncertain appearance altering conditions such as alopecia (Cartwright et al, 2008).

Young people's illness perceptions in the current research were found to be associated with their body esteem; greater perceived threat from NF1 was related to lower body esteem. However, both young people's and parents' perceptions were both found to be highly varied, on most items respondents scored across the range of scores, highlighting the variability of NF1 on all dimensions. The variance in responses underlines previous research which has suggested there is a lack of common identity for people with NF1 (Carrieri, 2011; Ablon, 1999). Within this varied experience, many respondents reported high levels of concern, and very little control over NF1. In interviews this lack of control had been discussed by all participant groups in terms of the uncertainty of the condition and not knowing how and when appearance could change. Managing uncertainty has been identified as a particular challenge of living with chronic conditions generally (Mishel, 1999), NF1 specifically (eg Ablon, 1999; Mouridsen & Sorensen, 1995; Rubenstein & Korf, 1990; Wellman, 1990) and is also important within adjusting to a visible difference (Rumsey et al, 2010). The lack of control evidenced in surveys and uncertainty described by young people and their parents in interviews may present a significant risk factor to positive adjustment to NF1.

The way in which the severity or noticeability of a visible difference impacts on an individual's adjustment and management of a difference is complex (Thompson & Kent, 2001). Clinical severity or the objective assessment of the noticeability of a visible difference has been found to have little connection with psychological adjustment (Bradbury, 1996; Macgregor, 1990), but rather individual subjective experience has been highlighted as important within adjustment (Ong, 2007; Moss, 2005). Interview findings appeared to point to little correspondence between the noticeability of young people's conditions and how they felt about their appearance. Health professionals reported that there was not a particular relationship between how an individual looks and how they feel about their appearance. However, parents talked of how visible different tumors and cafe au lait marks were and discussed how this impacted on their child. Subjective accounts of noticeability, from parents and young people, were therefore sought and investigated in the survey.

Findings from the surveys suggest that the noticeability of NF1 was important within parents' reports of their child's experience but was not an important aspect within young people's direct reports. Differences between parents' and young people's perceptions of the impact of severity of NF1 have been reported previously (Sebold et al, 2004; Counterman et al, 1995). Sebold suggests that these differences relate to young people's changing cognitive ability and point to their finding that older adolescents' scores became closer to their parents' assessments of severity. In this current research young people were substantially older (survey mean age = 20.4 years) than both Counterman's and Sebold's adolescent groups (mean ages = 11.8/15 years respectively) yet the differing importance of noticeability between parents and young people was still apparent. It is unclear exactly why parents reported noticeability as important. However reviewing interview findings suggests that it may relate to vigilance and concerns over how visible differences could impact on a child's life. During interviews some parents discussed their concerns that NF1 could become more visible over time, their concerns primarily related to how this could impact on their child socially. Parents described how these concerns led to them monitoring their child's appearance; they described watching for signs of NF1.

This has previously been described by Thompson and Kent (2001) who suggest such behaviour could lead to an increase in the emphasis placed on appearance by parents. Health professionals also commented on this and felt that parents may be very sensitive to changes to their child's condition. Managing uncertainty has been highlighted as a central aspect of parents' experiences of parenting a child with a chronic health condition (Stewart & Mishel, 2000) and vigilance has been described as a coping mechanism that parents use to manage this uncertainty (Jessop & Stein, 1985). This increased vigilance and awareness of appearance reported in interviews may lead to parents focusing on and emphasising noticeability within reports of their child's experience.

That young people's reports of noticeability were not significant within their happiness, social interactions or illness perceptions overall contradicts previous research with adults with NF1 which has pointed to a link between their reports of the visibility of NF1 and psychological wellbeing (Granstrom et al, 2012; Wolkenstein, 2009; 2001). However, it is important to note that while Granstrom et al report that adults in their study had a negative body image and visibility of the condition impacted on a range of measures including quality of life, they also report that body experience (defined as how secure and confident people felt about their bodies) mediated the relationship between visibility and psychological stress. Similarly, Lawrence et al (2006a), reporting on a study with burns survivors, found that the importance placed on appearance by an individual moderated the relationship between subjectively reported severity and their body esteem. These findings highlight that the importance placed on appearance and experience of appearance generally may be particularly salient aspects of living with a visible difference. This emphasises the importance of attending to general appearance evaluations for people with a visible difference, rather than noticeability.

The importance of appearance, regardless of noticeability, within young people's experience of NF1 is clear. Previous research has suggested that the salience of appearance may be an important factor in managing an altered appearance (Lawrence 2006a). An increased focus on appearance may lead to processing biases and may be linked to poor adjustment (Moss & Carr 2004). Young people's appearance evaluations were found to be an important factor within their wellbeing, as they are for many young people (Mendelson, 1997). Body esteem was the best predictor of young people's happiness and was associated with social comfort, stigma perceptions and illness perceptions for young people. Research suggests that during adolescence and into adulthood the awareness and importance of appearance increases (Moss & Rosser, 2008). Messages regarding appearance come from a variety of sources and affect young people in different ways (Cash & Smolak, 2012). As such it is difficult to predict who will or will not have appearance concerns during adolescence, either for those with NF1 or the general population. Findings in this thesis demonstrate that while body esteem was important to young people, it was not necessarily negative. Rather than seeing the experience of young people with NF1 as separate to the general population, it is important to view them first and foremost as young people and to remember that young people with a visible difference may have appearance concerns unrelated to a visible difference. As evidenced in interviews, any visible difference is a part of their overall appearance. Young people with a condition, such as NF1, might

still be prone to the appearance concerns faced by other young people such as weight, height, hair or may have no appearance concerns at all. It is essential that young people's experience of a visible difference is not assumed to be negative and pathologised (Wallace et al, 2007).

Being aware of and attending to the changing needs and levels of understanding of young people with a chronic health condition is an important aspect of working with young people (Sawyer & Aroni, 2005). Adolescence is identified as a time of increased appearance awareness (Moss & Carr 2004) and feeling well informed during this time of changing understandings may support the development of positive perceptions and may be empowering (Petrie & Weinman, 2006). All participant groups highlighted the importance of both good quality information and access to specialists (as an information source) as supportive of young people's experience and understanding of NF1, including management of the appearance related aspects of NF1. While coping patterns were found to be highly varied between parents, within this understanding NF1 and access to specialists and information were identified as particularly useful strategies, in line with previous research with other groups of parents (Cavallo, 2009; Fisher, 2001). Many parents described seeking information regarding NF1 when their child was initially diagnosed. However diagnosis may take place when a child is very young and there may be many developments in knowledge and treatment of NF1 over the years, as such over time the available information may change. Furthermore, as the developmental needs of a child change, so do the information needs of a parent (Hummelink & Pollock, 2006). Parents and health professionals reported that parents may require support and advice in managing their child's appearance related concerns. These concerns may not be apparent when a child is first diagnosed and may become increasingly important through adolescence (Ferner et al, 2007; Moss & Carr, 2004; Wellman, 1990). Survey findings demonstrated that the type of concerns parents held changed overtime. Initial concerns related to the medical aspects of NF1; however over time these changed and became more related to psychosocial concerns. This may mean that appearance concerns are not raised, and thus not addressed at initial diagnosis and may need to be revisited at a later point in a young person's life, with both parents and young people, reflecting the changing importance of appearance within young people's experience.

Findings suggest that appearance plays an important role within some young people's experiences of NF1. However the impact is highly varied. In line with previous research and reports by individuals with a visible difference, individual perceptions are clearly important within adaptation (Rumsey & Harcourt, 2012; Lansdown et al, 1997). While parents feel the noticeability of NF1 is an important factor within young people's experience, this was not the case for young people themselves. For young people, appearance concerns may relate to anxiety regarding the uncertainty of future changes and a perceived lack of control over these changes, rather than the noticeability of NF1. Findings particularly highlight the varied and subjective manner in which young people manage NF1 during adolescence.

### 9.2.2 Social Interactions

The uncertainty and lack of control related to NF1 discussed in the previous section, coupled with the importance of general appearance evaluations within young people's experiences as well as parental emphasis on noticeability, could lead to what Thompson and Kent (2001) refer to as 'appearance consciousness'. This heightened awareness of appearance may be a risk to positive adjustment and resilience, particularly with regards to its impact on social interactions and perceptions of stigma. Wallace et al (2007) report that the value placed on appearance prior to changes was relevant within accounts of resilience within their research with young women with appearance changes resulting from cancer treatment. Findings presented in this thesis have highlighted the importance of appearance evaluations within social interactions, perceptions of NF1 and young people's happiness.

How appearance consciousness may impact on social experience is explained by Moss and Carr (2004). They suggest that if individuals are very aware of their appearance and have concerns over how others may react, then they may be more likely to interpret social experiences negatively, attributing any challenges to their visible difference. Certainly the impact of NF1 on young people's day-to-day experiences was generally described in terms of how it affected social interactions. In interviews a common assessment was that the appearance related aspects of NF1 were not an issue for the young person as an individual, but rather any difficulties were related to how others responded to the way they looked.

The importance of the social world when living with a visible difference is well documented within the appearance literature (eg Rumsey & Harcourt, 2012; Feragen, 2010; Thompson & Kent, 2001; Newell, 2000). Social interactions can be challenging for young people with a visible difference (Lovegrove & Rumsey, 2005) but friendships and social support can also be an important factor in resilience (Williamson et al, 2010). If young people do not feel socially confident and have concerns relating to how others perceive them, then this is likely to impact on day-to-day social relationships (Moss & Carr, 2004; Kent & Keohane, 2001). Young people with NF1 have previously been reported as having lowered social skills and competence than peers (eg Johnson et al, 1999; North et al, 1997; Dilts et al, 1996). It has been suggested that commonly associated learning and behavioural difficulties (including ADHD and ASD) may explain this for some young people (Barton & North, 2004). However it is also worth bearing in mind that young people with NF1 may have attention deficits and difficulties recognising social signals separate to any specific difficulties (Isenberg et al, 2013; Huijbreghts et al, 2010a b).

The findings presented in this thesis suggest that young people's perceptions of stigma and how comfortable they feel socially are important factors. In line with previous research, young people's happiness was found to be related to feeling socially confident and perceiving lower levels of stigma (Rumsey & Harcourt, 2005; Moss & Carr, 2004; Thompson et al 2002; Thompson & Kent, 2001; Kent and Keohane, 2001; Newell, 2000). How social encounters are viewed and managed was clearly important for young people, in terms of their wellbeing and also their appearance evaluations. Young people, parents and health professionals all described varied social experiences and, similarly to

previous reports (Rumsey et al, 2004; Lansdown et al, 1997) concerns related to managing other people's reactions. Young people and parents explained that staring from strangers and questions about their appearance were particular challenges. For some, these interactions were brushed off, some people were happy to explain NF1 and what any visible marks were. Others however felt that such encounters were intrusive and upsetting. A particular challenge described by all participant groups was a perceived lack of knowledge of NF1 by the public generally. Many participants (including health professionals) felt that the condition was poorly known and often misunderstood and that this contributed to young people's anxiety related to discussing the appearance related aspects of the condition. Participants described how visible differences such as tumors and cafe au lait marks were hard to explain as a great deal of explanation of their condition was needed. This was described, by some, as leading to fear and concern about being asked questions which in turn was described as leading to a range of behaviours including avoidance of social situations, covering visible marks or, as a few people described, inventing alternative narratives to explain their appearance. This suggests that young people and their parents may benefit from support in managing the reactions of others and preparing ways of managing possible comments.

Negative experiences may lead to a greater awareness of any visible difference and feeling self-conscious. Concerns about others' negative reactions can lead to social anxiety (Lovegrove & Rumsey, 2005) leading to a desire to avoid such situations; this can then impact on the development of social skills and lower levels of self confidence which in turn can increase social difficulties (Feragen et al, 2010; Chamlin, 2006; Tan, 2004). Some young people and parents talked of avoiding social situations; the possible impact of this on social skills is explained by Newell's (1999) fear avoidance model. This model suggests that if a person manages social anxiety by avoiding social situations, they may feel this behaviour is helpful, avoiding social situations may therefore become a strategy to manage social anxiety. Kent & Keohane (2001) suggest that this may lead to an increased likelihood of people attributing difficult situations to their appearance, which in turn increases the importance of appearance within this experience. This may be compounded for young people with NF1 due to the lowered social skills reported as associated with the condition (eg Sebold et al, 2004; Dilts et al, 1996; Benjamin et al, 1993) as well as the reported difficulty in explaining the condition, due to a lack of public awareness of neurofibromatosis.

In addition to managing the reactions of the general public, the appearance related aspects of NF1 may impact on friendships. NF1 has been found to have a profound impact on social aspects of quality of life (Krab, 2008). Children with NF1 have been found to have fewer friends and difficulties socialising (Noll et al, 2007) and recent research with adults with NF1 has found that these difficulties continue into adulthood (Pride et al, 2013). Social experience has been described as supportive of positive adaptation to chronic illness (Reiter-Purtill & Noll, 2003; La Greca, 1990) and all participant groups in this research programme highlighted in interviews that adolescence was a time during which social aspects of life become more important. This increasing importance of friendships and the social sphere within young people's experiences of a visible difference has previously been highlighted by researchers, many of whom have suggested this may be an arena in which resilience

can be supported (Williamson et al, 2010; Fox et al, 2007; Wallace et al, 2007 a & b). Several participants in the current research described how teasing and negative social experiences first occurred following the transition to secondary school. Health professionals and parents reported changes in social experiences as young people went through adolescence. However, although qualitative findings pointed to a change in social experiences no significant differences were found on the measures of social comfort or stigma in relation to participants' age.

The relationship between young people's social interactions and illness perceptions, appearance evaluations and wellbeing highlights the complexity and many different factors that can impact on the day-to-day lives of young people with NF1. Findings presented in this thesis demonstrate that how confident and happy young people felt socially is likely to be influenced by how they perceive their condition, how they feel about their appearance, how happy they are and vice versa. The previous section considered how illness perceptions might relate to individual processes and appearance evaluations. It is also important to recognise the role of illness perceptions in social interactions. If young people have negative perceptions of their condition this is likely to impact on how socially confident they feel; they may feel anxious about explaining their condition and are likely to have greater concerns about how they look and how others might respond. Conversely negative social interactions are likely to impact on their perceptions of NF1. Findings presented in this thesis demonstrate that illness perceptions, while highly varied, are clearly important across young people's experiences of NF1.

This section has considered how a possible heightened awareness of appearance may lead to social challenges for young people with NF1. Findings indicate that illness perceptions generally as well as perceptions of stigma and social comfort are important within wellbeing, although they are not necessarily negative. Social challenges appear to relate to worries regarding how others might react and concerns about explaining the condition rather than regular teasing and negative experiences.

### **9.2.3 Section Conclusion**

This is the only known research that has explored the role of appearance within young people's experience of NF1. The findings presented in this thesis suggest that while young people are likely to perceive NF1 in many diverse ways, how they feel about their appearance, manage uncertainty and social interactions are key factors. In line with previous literature regarding the management of an altered appearance (including Rumsey & Harcourt, 2012; Thompson & Kent, 2001) this thesis emphasises that the role of appearance within young people's experiences of NF1 relates to both individual processes and the social context. Self perceptions of appearance are highly salient within adjustment to an altered appearance (Moss & Rosser, 2012) and have been identified in this current research as particularly relevant within the happiness of young people with NF1. The importance of, and relationship between, the social interactions of young people and their perceptions of NF1 is a new finding and one in need of further exploration. In particular the reported uncertainty and low levels of control, in addition to changes in understanding their condition across adolescence, may

mean that young people with NF1 face particular difficulties in adjusting to and managing their condition through adolescence into adulthood. While health professionals and parents appear to concur with young people's reports of much of the day-to-day impact of NF1, a notable difference between parents and young people lies in their interpretation of the importance of the noticeability of NF1.

Adapting to, and living with, a visibly different appearance has been described as an evolving process (Prior, 2009), managing a changeable unpredictable appearance may be particularly challenging (Rumsey et al, 2010). Yet, young people with NF1 do not report particularly low levels of happiness, appearance evaluations or negative social interactions. In line with findings with young adults with other genetic conditions, such as Marfan syndrome (Tongerloo & Paepe, 1998), many young people with NF1 are happy and feel positive about social interactions and their appearance. As such, and due to the highly varied accounts of experience of NF1, supporting families and young people to be resilient and happy against a backdrop of uncertainty may be particularly beneficial for young people with NF1.

### **9.3 Application of findings**

The aim of this thesis is to explore and describe the role that appearance plays within young people's experiences of NF1 in order to provide a basis on which to inform support and healthcare. Having described the role of appearance and considered how it impacts on young people in the previous section, this current section moves onto the second part of this aim and considers how findings might be applied in order to support young people with NF1.

Adolescence and early adulthood is a time during which health interventions can be especially effective and positive coping and resilience can be supported and developed (Ahern et al, 2008; Holmbeck, 2002). Qualitative findings presented in this thesis highlight that adolescence is a time during which young people may reconceptualise their condition. As such, this developmental stage could be a time of opportunity during which interventions and supportive care may be able to promote positive management of the appearance related aspects of NF1 for the rest of an individual's life. Current recommendations for the care and treatment of individuals with NF1 (including Hersh, 2008; Ferner et al, 2007; Korf, 2001; North et al, 1997; Huson, 1989) highlight the importance of accurate information and psychological support for parents, children and young people. The Clinical advisory board of the Neuro Foundation (Ferner et al, 2007) specify that management of NF1 in the UK should include referral to a clinician who has specialist knowledge of the condition if a diagnosis of NF1 is being considered. The consensus statement also specifically highlights the importance of educational and psychological support for young people with NF1. The following sections specifically consider how aspects related to appearance could be addressed within these recommendations.

Rumsey and Harcourt (2012) highlight the importance of ensuring that appearance research is applicable. In order to consider the applicability of findings the CAR framework of appearance related

interventions was considered (Rumsey & Harcourt, 2012: figure 12 below). This framework illustrates how support for people with a visible difference can be offered at differing levels of intensity. The framework is a tiered model of intervention, as the pyramid rises the intensity of intervention increases and the number of individuals requiring the intervention lessens.

Societal campaigns (level 0) might aim to alter the way in which people view visible difference or encourage variation in appearance ideals. Current campaigns in this area are organised by groups such as Changing Faces who campaign on behalf of those with a visible difference. Their 'Face Equality' campaign aims to promote fair and equal treatment for people with a visible difference, raising public awareness and challenging negative attitudes (Partridge, 2012). Examples of level 1 interventions include targeted information delivered within a health care setting, and peer support such as online discussion forums. Level 2 interventions are aimed to be accessed independently and might include self help leaflets or online support. Interventions at this level address particular concerns such as social skills and should be easily accessible and designed to stand alone. Level 3 interventions may address similar issues as level 2 but would be more intense and would involve trained professionals from support groups or health professionals. Level 4 would involve brief specialist led counselling whilst level 5, face to face specialist led interventions, would be considered for those experiencing very high levels of distress.

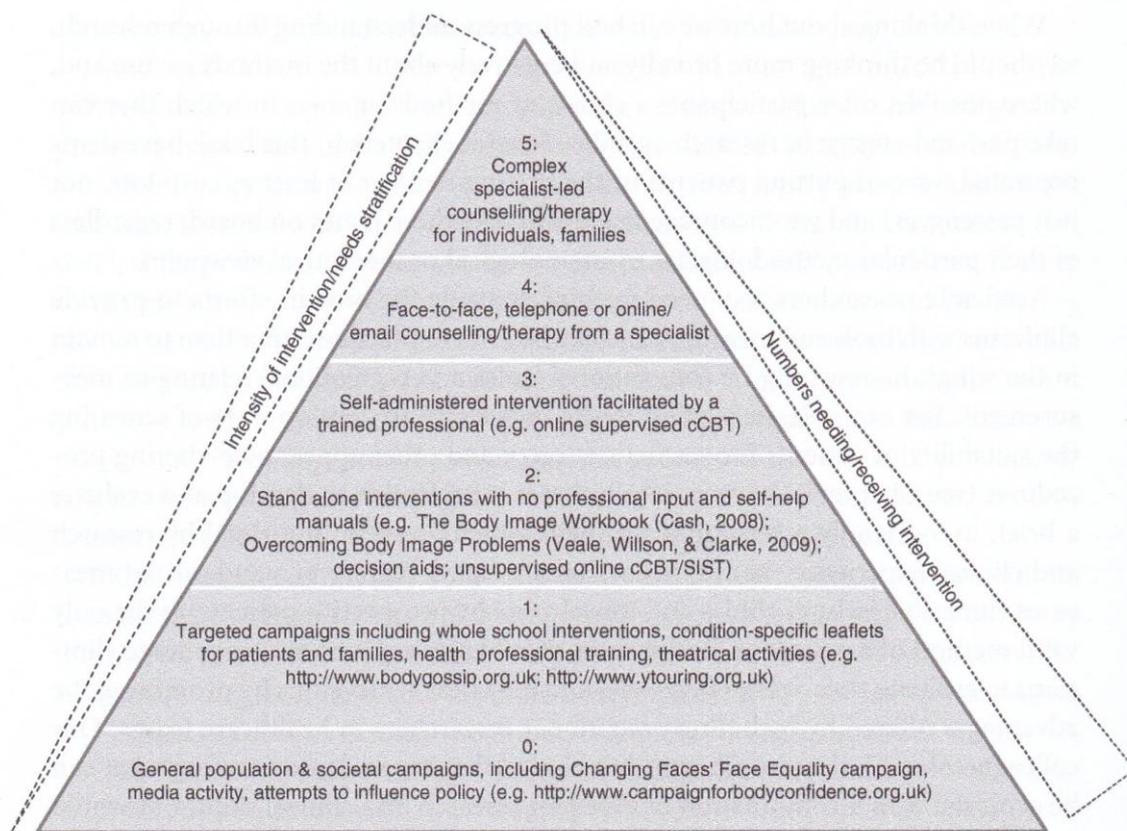


Figure 12. The CAR framework of psychosocial interventions (This material was originally published in The Oxford Handbook of The Psychology of Appearance edited by Rumsey N and Harcourt D and has been reproduced by permission of Oxford University Press [<http://ukcatalogue.oup.com/>]. For permission to reuse this material, please visit <http://www.oup.co.uk/academic/rights/permissions>)

Findings presented across this thesis suggest that while the impact of NF1 is highly varied, managing other people's reactions to their appearance, engaging with healthcare, seeing specialists who understand NF1 and being able to access information and support regarding NF1 and appearance throughout childhood and adolescence was important to both young people and their parents and this was acknowledged by health professionals. The following sections consider these different aspects within the framework of the CAR pyramid (Figure 12). Section 9.3.1 briefly considers how societal campaigns might be supportive of young people's experiences of NF1. Jenkinson (2012) suggests that interventions addressing information provision (level one of the CAR pyramid), should be offered to all people with a visible difference. Therefore section 9.3.2 considers how findings could be applied in relation to supporting understanding and information provision for parents and young people, and asks how this might impact on the provision of healthcare for young people with NF1. The day-to-day impact of NF1 was described by many primarily in terms of its impact on social interactions; therefore how findings could be applied in order to support young people's social skills and interactions at levels 2 and 3 is discussed in section 9.3.3. Section 9.3.4 then considers interventions offering more intense therapy for individuals with complex social needs at levels 4 and 5.

### **9.3.1 Level 0: Societal campaigns**

Rumsey and Harcourt (2012) highlight the importance of ensuring that appearance research promotes appearance satisfaction for individuals (with or without a visible difference) whilst also encouraging societal change so that acceptance of a range of diverse appearances is the norm. On a societal level this relates to challenging and changing perceptions of visible difference (Lansdown et al, 1997). It is important to recognise that the impact of managing a visible difference on young people generally, and NF1 specifically, occurs within a socio-cultural environment in which appearance is highly valued (Rumsey & Harcourt, 2012). The current research demonstrates that how young people with NF1 feel about their appearance is highly significant to them, as it is for many young people with and without visible differences (Rumsey & Harcourt, 2012; Lawrence et al, 2006a; Cash, 2004). Both individual societal interactions and the wider social landscape were found to be important to young people with NF1.

The importance of young people's social interactions is highlighted throughout this thesis and appears to be a particularly critical area in which to support young people with NF1. However, it is important to recognise that social interactions are a two way process. While supporting the development of young people's social skills is likely to be supportive of their experience (Jenkinson, 2012), research demonstrates that they are not imagining social challenges (Hearst, 2007). For instance children have been found to be less willing to interact with children with a facial difference than one without (Masnari et al, 2013) and 9 out of 10 people surveyed by Changing Faces (Changing Faces, 2013) were found to hold negative attitudes towards people with a visible difference.

In terms of NF1, any perceived stigma may be compounded by the additional stigma associated with genetic conditions (Peters et al, 2005) as well as the perceived lack of public awareness of the

condition reported in the current research. In interviews, almost all participants felt that managing NF1, particularly the appearance related aspects, would be easier if others knew about the condition. Furthermore, young people in the current research described how persistent (inaccurate) links between NF1 and 'the elephant man' made them reluctant to tell others the name of their condition. That this link persists was evidenced during the course of this research. In 2012, a young person with NF1 appeared on the UK television programme 'This Morning' to talk about her condition. She was billed as 'the elephant girl' prompting much anger and distress from many people with NF1, who organised themselves through online groups and called ITV to complain about the billing, which was subsequently altered. However, although the presenter acknowledged that people had complained he also reiterated the link with 'the elephant man' and suggested the link was disputed.

Many participants in this research commented that NF1 is not a particularly rare condition; they explained that it is roughly as common as Down's syndrome and cystic fibrosis, both of which people felt were better known amongst the public. Raising awareness of NF1 is a priority for The Neuro Foundation and its members, however addressing this is likely to be challenging. The perceived lack of recognition of the condition may be partly explained by the variability of NF1. The condition presents in many diverse ways. The lack of a core NF1 identity has been highlighted previously (Carrieri, 2011; Ablon, 1999) and this may challenge how recognisable it is to others. Large scale publicity campaigns are costly and educating the wider public about NF1 in particular and visible difference in general is likely to be a gradual process. Therefore a focus on supporting young people to positively manage social interactions against a backdrop in which appearance is highly valued and NF1 not widely recognised is essential.

### **9.3.2 Level 1: Understanding NF1 and information provision**

Current UK and international guidance (including Hersh, 2008; Ferner, 2007; Korf, 2001) recognises the importance of attending to the educational and psychosocial needs of young people with NF1. This thesis highlights the need to address issues relating to appearance within this. Parents and young people described an increasing awareness of the appearance related aspects of NF1 through adolescence. It is therefore important that health professionals working with young people with NF1 are aware that appearance is important within wellbeing. It is important that they understand that this is not necessarily associated with the noticeability of the condition, and the day to day impact often relates to young people's social interactions. The health professionals surveyed demonstrated a high degree of awareness of these issues that some young people with NF1 and their families face and recognised the complex role that appearance plays within this. However, the health professionals in the current research were primarily specialists, many were genetic counsellors, and may not be representative of whom young people actually see, particularly following transition to adult care. Young people and parents reported seeing a range of different health professionals, both specialists and non-specialists who may not be as knowledgeable as those surveyed in this research. Certainly, common concerns for parents and young people related to whether non specialist health professionals fully understand NF1.

Findings from both interviews and surveys suggest that having an opportunity to engage with a health professional with specialist knowledge of their condition may be highly desirable for young people with NF1. This need was described in terms of supporting young people's informational needs, putting information into context and addressing any appearance related concerns; all of which are likely to inform young people's illness perceptions which were found to impact across experience. Young people with NF1 describe uncertainty as a particular challenge and being able to talk to health professionals who can address this is likely to be highly valued and highly supportive of developing positive illness perceptions. In interviews, health professionals reported that an important aspect of their role was putting information into context, both for parents and young people. Information provision and supporting understanding has been found to support the development of autonomy which is crucial within young people's adaptation to a chronic illness (Spear & Kulbok, 2004) yet previous research with adults with NF1 has identified that patients are not happy with information provided for them (Dheensa & Williams, 2009). Information can be important in normalising young people's experience. It is also essential to be aware that for many young people learning about genetic conditions happens throughout their childhood and adolescence (Gallo et al, 2005), learning about NF1 is a result of many events and sources of information over many years. The communication of genetic information generally often occurs within families and is complex; families differ in their management of genetic information (Gaff et al, 2007; Forrest et al 2003). As young people are likely to learn about their condition from their parents, support in communicating information may be required (Metcalf et al, 2008). As such it is imperative that parents are provided with up to date information and support regarding the condition not only for their own benefit but also to support them in supporting their child's understanding (Metcalf et al, 2011).

It is important to recognise that in the current research some young people described a preference to learn about NF1 outside of the family for a variety of reasons. While some young people reported that their parents were the primary source of information and support regarding NF1, others described their parents as knowing little about the condition or not wanting to discuss it. Particularly if a parent is affected by NF1 themselves, their own direct experience is likely to impact on if and how they discuss the condition (Clarke et al, 2011). While research has highlighted the role of families in communicating genetic information, interview findings also underline the importance of not assuming that parents will have informed their child or accurately informed their child about the condition or that young people want to learn about NF1 from parents.

During adolescence, young people may value health professionals who ask them if they have appearance worries and take their appearance concerns seriously, providing advice or referring them for specialist support (Williamson & Wallace, 2012). A first step in adjusting to an altered appearance, at any age, can be in acknowledging and legitimising the concern (Clarke, 1999). In addressing issues around appearance, and simply asking about appearance, health professionals can legitimise and normalise the concerns of individuals. An opportunity of this kind could be provided through

genetic counselling. Previous research has suggested that young people with a genetic condition may have gaps in their knowledge and would like to receive genetic information, through genetic counselling, at a younger age than they are generally offered (Szybowska et al, 2007). Genetic counselling is more specialist than information provision and therefore genetic counsellors may be particularly well placed not only to address information provision at level 1 but also to support young people (and parents) in adaptation and accessing higher levels of support. However, it is important to note that genetic counsellors may need training and support so they are able to provide advice related to appearance concerns or refer on to appropriate support. Training, on appearance related issues, including study days or longer courses, has been found to be effective but requires both time and resources (Clarke & Cooper, 2001).

The important role that genetic counsellors could play within young people's adaptation to NF1 is not a new suggestion. Huson et al (1989) highlighted that very few families (7%) in a study of families with NF1 in South Wales had received genetic counselling. She suggests that genetic counselling could support young adults, whether they have specific problems or not. Gaff and Clarke (2006) also highlighted the important role that genetic counsellors can play in supporting adjustment to visible difference and the stigma experiences of individuals with NF1, including during adolescence. However, most young people in the current research did not report having received genetic counselling, although some parents reported talking to a genetic counsellor at initial diagnosis. In the case of genetic conditions such as NF1, it may be appropriate to offer access to young people at key developmental times, such as transition to adult health care. Not all young people would desire or require such contact; however findings in the current research suggest that the opportunity to discuss the condition and any concerns, including appearance concerns, may be highly welcomed by many young people. Offering genetic counselling to young people as they transition to adult health care may be highly beneficial, not only in terms of supporting adaptation, and acknowledging psychosocial concerns, including appearance, but also in terms of information provision. While it was not a focus of this thesis, findings suggest that young people are concerned about the possible impact of NF1 on having children yet this was not acknowledged by health professionals. Genetic counselling would be an appropriate forum in which to discuss these concerns and gain information on inheritance. The feasibility of offering genetic counselling to young people during transition would need to be evaluated in terms of accessibility, cost-effectiveness, timing, impact upon genetic counselling services and patient satisfaction.

It is important to note that while many young people and parents desired access to specialists, many of the (mainly specialist) health professionals in this programme of research felt that young people with NF1 were difficult to engage with. Similar findings have been reported with other young people with genetic conditions and engaging adolescents in genetic counselling is an important issue (Tse et al, 2013). Therefore, how genetic counselling is offered to young people may need some consideration. One way of addressing this challenge may be in considering the use of innovative methods. For instance being able to access specialists through email may be useful for young people. Harvey et al (2008) explored the use of email between young people and health professionals and

found that the medium was one in which young people were very adept and engaged with. In particular they point out that health consultations and asking questions can be challenging for young people in a regular health setting, email allows young people to ask difficult or sensitive questions. This may be particularly supportive for young people with a condition such as NF1 as it provides a comfortable arena in which to address sensitive concerns including appearance concerns. Using online methods with young people has been identified as an advantageous way for health professionals, such as genetic counsellors, to engage with young people (Skinner et al, 2003).

The desire to access specialists was discussed by young people in terms of being able to see health professionals who understood NF1 clinically, and also as an information resource. Therefore another way in which to engage with young people may be through online events. For instance The Neuro Foundation has previously supported online 'meet the expert' events where a specialist health professional has been available online at a specified time to 'meet' people and discuss NF1 queries. Sessions in this manner aimed specifically at young people run by specialists may be an avenue to consider and explore and evaluate further. In addition to accessing specialists, whether Neuro Foundation specialist advisors, specialist health professionals or genetic counsellors, finding information about NF1 was highlighted as challenging by young people.

All participant groups in the current research reported that young people with NF1 often used the internet as a health information resource. But as previously reported with other population groups (Cline & Haynes, 2001), participants were concerned about the quality of information available. A particular concern raised was the pathologising and exaggerated images used of NF1 symptoms on some websites and the appearance related concerns these might or did cause. Findings that health professionals had concerns about the reliability of information on the internet suggest that having a reliable online information resource for young people about NF1 may be highly beneficial. Findings presented in this thesis suggest any information should address issues around appearance and social interactions, in particular talking about NF1. This could be developed in association with support groups such as The Neuro Foundation in the UK who part fund a number of specialist advisors who work to provide information and support for people with NF1 and recognise the importance of specialist information provision. Developing materials specifically for young people would need funding and materials would need to be evaluated. Developing and maintaining a relevant information resource on the internet may be challenging, particularly in terms of management and relevance (Newman et al, 2009). However, the current findings point to the internet being the most commonly accessed site of information regarding NF1 for the young people surveyed and 85% of health professionals indicated a need for online support. Furthermore, previous research suggests that the internet may be the primary health information resource for young people (Gray et al, 2005). However, online materials and events should not be the only information provision for young people with NF1. While many young people use the internet widely (Lee, 2008), not all young people have access to the internet and many, including those with specific learning difficulties, may find accessing the internet challenging (Gui & Argentin, 2011). It is also important to bear in mind that some young

people may develop appearance concerns after reading online materials or have concerns that are not met with such a resource. In such cases young people need to have access to additional support.

In addition to supporting young people directly, this thesis highlights the importance of supporting the diverse and changing informational needs of parents. Parents can be highly influential in their child's adaptation to an altered appearance throughout childhood, adolescence and into adulthood (Bellew, 2012). As such, supporting their informational needs is likely to indirectly support young people. Across this thesis and in line with previous research (Hummelinck & Pollock, 2006) parents described their concerns and information needs changing alongside their child's development. Early concerns were described as medical and these changed over time, becoming more social and emotional and included managing appearance concerns and social situations. Parents wanted to support their child in managing any challenges they were facing but also reported a desire to pre-empt concerns and put measures into place to support their child in advance of concerns arising. Parents may therefore benefit from support and information regarding NF1 and appearance in advance of adolescence, advising them how they can support their child through this stage. There are a variety of strategies that parents could put in place for their child. Some parents felt this could be accomplished through ensuring children had a strong and varied social life and others talked of how the family modelled positive conversations about appearance, parents also described normalising concerns. However whilst some parents described using such strategies it may be that many families would benefit from advice on this subject. For some parents support may be provided through meeting other parents either in person, or online. Such information and advice could also be delivered more formally through support groups such as The Neuro Foundation and information should be available through NHS clinics for those who are not involved with support groups. Further research may be required in order to develop such information, however findings presented in this thesis suggest that parents would welcome advice regarding how to support their child across adolescence and within this an important message to feedback to parents is the reported lack of a relationship between noticeability of NF1 and wellbeing for young people.

NF1 has been described as particularly challenging due to its lack of a clear identity and the diversity of experiences (Carrieri, 2011; Ferner, 2007; Ablon, 1999). In the current research, young people and parents describe feeling a lack of control and a great deal of uncertainty, they were also found to hold highly diverse illness perceptions, which related to their wellbeing, appearance evaluations and social interactions. Therefore ensuring clear and accurate information is provided for young people and parents is essential, in order to support the development of positive illness perceptions and a clear understanding of the condition. However, as highlighted in this section, a particular challenge lies in how information should be provided and by whom and further research is needed to explore this and then to subsequently evaluate the impact of information provision, this is considered further in section 9.5.

### 9.3.3 Level 2 and 3: Social interactions

It is important to emphasise that the current research demonstrates that not all young people will find NF1 a challenge or have appearance concerns; many were happy and managed well. However, in line with previous research with adults with NF1 (Pride et al, 2013), the particular challenges that young people with NF1 reported generally related to the reactions, or concerns about the possible reactions, of other people. Managing the impact of a visible difference, on an individual level, has been described as relating to being able to positively manage social interactions (Thompson & Broom, 2009). This could be addressed through interventions designed to support young people's management of social interactions at different levels of intensity, depending on individual need. Level 2 interventions would be designed to be accessed independently (for instance workbooks on how to talk about NF1, or independently accessed online social skills interventions) while level 3 interventions would be more intensive and would be facilitated by trained professionals.

A systematic review of interventions for people with a visible difference concluded that cognitive behavioural therapy (CBT) and social skills interventions were the most successful types of intervention (Jenkinson, 2012). Social skills have been identified as important in management of a chronic illness (Reiter-Purtill & Noll, 2003). Research with adults with NF1 has identified them as demonstrating less prosocial behaviour than unaffected adults (Pride et al, 2013) and young people with NF1 are identified as having poorer social skills than peers (Sebold et al, 2004; Johnson et al, 1999; Dilts et al, 1996; Benjamin et al, 1993). As outlined in Chapters 1 and 2, young people with NF1 may have associated learning difficulties and incidences of ASD and ADHD are high. Taking all of these findings into consideration, social skills interventions may be particularly beneficial.

Previous research with young people with NF1 suggests that such interventions could include components related to recognising social signals (Huijbregts et al; 2010 & 2011) and recognition of gestures and tone of voice (Lehtonen, 2013) while the current research highlights the need for support in talking about NF1 and managing others' reactions. As in the delivery of information, a particular challenge may be in how to deliver social skills interventions to a geographically diverse group of individuals who are cared for in many different settings. Again, the answer may be in using online methods. Online CBT and social skills support has been found to benefit adults with a visible difference through reducing anxiety, depression and appearance concerns (Bessell et al, 2012), and CBT approaches have been demonstrated as effective in supporting young people with a visible difference (Jenkinson, 2012). An online intervention for young people with visible difference, developed by the Centre for Appearance Research is under evaluation (YP Face it, 2013). As this intervention is specifically developed to support young people with a visible difference this may be of interest to professionals working with young people with NF1, this is designed to be accessed at either level 2 or 3 of the CAR framework. Online provision may be appealing for young people with NF1 in terms of being able to access support at convenient times and also without the need to travel. Health professionals in the current research identified that engaging young people and arranging

sessions in clinics could be difficult due to young people being in geographically diverse locations and also those aged under 16 years old in particular are often reliant on parents for travel.

While online interventions, with or without the support of a trained professional, may be desirable for many young people, others may welcome the opportunity to develop their social skills in a group setting and meet others of their own age with the same condition. Several young people in interviews discussed their desire to meet others with NF1 and talked of the shared understanding that was achieved through such meetings. However, providing this opportunity can be challenging particularly as young people with NF1 are cared for in diverse settings. This could be achieved through organising events specifically for young people. For instance the British Columbia NF association (BCNF, 2014) organises transition events for young people with NF between 16-30 years old in order to provide information relevant to the age group as well as the opportunity to meet others with the condition. While these have not been formally evaluated, feedback from attendees has been positive. In addition to information, events such as these could include the provision of self help guides and social skills interventions at level 2 as well as provide the opportunity for young people to meet specialist health professionals who may be able to signpost them towards additional support at a higher level of intensity if required. The content of such events could be developed in collaboration with young people with NF1 and the impact measured by exploring the number of young people who attend and asking for feedback from those who attend.

A meeting or day-long event may not provide the time to go beyond a low level of information provision and minimal self help. This may be highly beneficial to many young people and may provide all the support that they need. However some may benefit from more intensive provision. During interviews some young people mentioned having attended camps for young people with NF1, and discussed the positives of these experiences. Therapeutic summer camps could aim to increase NF1 related knowledge (at level 1) as well as deliver psychosocial interventions to improve social interactions skills (at levels 2 and 3) they may also provide an opportunity to identify individuals in need of additional support (at levels 4 and 5). Research suggests that camps can reduce feelings of isolation and increase self-esteem for individuals with a range of different conditions including burns injuries (Rimmer et al. 2007) and altered facial appearances (Tiemens et al. 2007). Furthermore, Bakker et al (2011) have demonstrated a modest improvement in young people's satisfaction with their appearance following attendance at a camp for young burns survivors. In his thesis, Allsop (2012) specifically explored summer camps run by the Children's Tumor Foundation in the USA for young people with neurofibromatosis type 1 and 2. These camps included therapeutic activities designed to strengthen social self efficacy and social skills and findings suggest that these activities were successful in increasing individual's social performance with peers. Maslow and Lobato (2010) highlight in their literature review of summer camps for children and young people with burns injuries that qualitative studies suggest that attending burns camps can decrease isolation, promote coping skills as well as improve self-esteem, and social skills. However, it has proved difficult to quantify how

any such improvement has occurred (Gaskell, 2007). The content of any interventions at camps for young people with NF1 would need to be evidenced based and evaluated.

It is also important to note that camps are costly, meaning that funds would have to be raised to ensure that all young people can access them. Furthermore the age at which young people are offered places on camps would also have to be considered carefully, parents of young people with a genetic condition have previously highlighted some concerns regarding their children's readiness for camps, particularly in terms of finding out information they may not be ready for (Plumridge et al, 2012). While there is some suggestion that residential camps may support social skills development, overall there is little evidence that group based social skills training for individuals with a visible difference is effective (Jenkinson 2012). Further research and rigorous evaluation of the effectiveness of social skills interventions within a residential camp setting are needed.

#### **9.3.4 Levels 4 and 5: Counselling and intensive psychological support**

The CAR framework suggests that counselling and specialist in-depth psychological support at levels 4 and 5 should be available to the minority of individuals who require more intensive psychological support and those who exhibit a high level of distress. A particular concern raised by participants in this current research was the accessibility of this type of psychological support. Many young people and parents described being unsure of how to access psychological support and many health professionals felt that such services were hard to access. For a tiered model of intervention to work effectively access to more in-depth and intensive psychological support needs to be realistic and health professionals, parents and young people need to understand routes into support.

Within the NHS in the UK, psychological support is not routinely offered to individuals with a condition that does or could cause a visible difference (Hanson & Clarke, 2009). Some psychological support at level 4 is provided by support groups (such as Changing Faces) and in the South West of England there is one specialist psychological service 'Outlook', which provides support for children, young people and adults with appearance concerns. The interventions and support delivered by the service has been found to be effective for children, young people and adults (Cadogan et al, 2006; Kleve et al 2002).

Hansen and Butler (2012) suggest that psychological care for people related to appearance should be embedded within local mental health services or within care teams. However the provision of psychological support for individuals with appearance concern has been found to be highly dependent on specific care teams (Clarke et al, 2003) meaning that increasing awareness of appearance related issues, may be an important action to support young people's access to more intense psychological support where needed. For instance Rumsey and Harcourt (2012) explain that when researching in general medical practices they found many family doctors were surprised by the number of patients who were interested in taking part in appearance research and reported appearance-related concerns. It has been suggested that the perpetuating myth that noticeability and psychological

distress are related might mean that those with less visible differences are less likely to be referred for specialist psychological support (Hansen & Butler, 2012). In relation to findings presented in this thesis, challenging myths regarding who is likely to be affected by appearance concerns with health professionals who work with young people with NF1 is essential.

Findings suggest that providing intensive psychological support for individuals who require short term counselling (at level 4) or more intensive therapy (at level 5) is likely to be highly valued by young people with NF1. The limited evidence that has explored psychological input for individuals with a visible difference suggests such input would be effective. However specialist counselling and psychological provision is costly and whether young people are referred for counselling may depend on the health professionals they see for their NF1 and their understanding of appearance related distress. While many young people with NF1 are happy and may not require counselling and intensive psychological input, it is important that services are available and accessible for those who do.

### **9.3.5 Section conclusion**

This section has considered how findings from across this thesis could be applied to support the care of young people with NF1. Specifically there is a need for access to specialists who are knowledgeable about both NF1 and issues around appearance. Secondly good quality information should address the changing informational needs of both young people and parents. Thirdly, young people may benefit from specific support negotiating the social aspects of NF1, in particular answering questions about NF1 and finally more intense psychological support should be available for young people who have significant appearance related distress.

However, developing and delivering these interventions would need further research and evaluation. For instance any information for young people and parents should be developed based on existing evidence and would need to be evaluated. Social skills interventions and access to psychological support across different levels of intensity needs to be available and accessible. Not only will this mean the development and testing of interventions but also the education of health professionals who work with young people with NF1. Research that is needed to bridge the gap between findings presented in this thesis and recommendations for care are considered in section 9.5.

The different interventions discussed in this section take the form of conversations with specialists in clinics and support groups, leaflets for parents and young people, specific self help guides, computerised social skills and CBT support or groups sessions and one-to-one intensive individual support. What is important is that young people's (and parents') changing and developing informational needs are met, and any specific support needs are addressed during adolescence in order to support long term resilience throughout adulthood. Within this it is imperative to bear in mind the highly diverse experiences of young people and the varied impact that the appearance related aspects of the condition can have.

## 9.4 Methods

This section evaluates the use of research methods across this thesis. A pragmatic approach was taken, as discussed in Chapter 3. Methods were chosen to meet the different aims and questions of the different stages of this programme of research, reflecting the researcher's belief that knowledge is both constructed and based on the reality of the world we experience and live in (Morgan, 2007). Methods should be employed fittingly and their use explained and justified (Creswell, 2003). This section reflects on and evaluates the methods used and the participants included in the programme of research and critically considers the value of a mixed methods approach.

There is much debate regarding what constitutes quality in mixed methods research and no clearly agreed criteria exists for evaluating such research (Creswell & Plano-Clark, 2007; Sale & Brazil, 2004). Given this lack of an evaluative framework, O'Cathain et al (2008) devised a set of questions in order to assess the quality of mixed methods studies in health services research (Good reporting of a mixed methods study, see below). These questions are now used as a framework in order to evaluate the methods used and participants included in this current programme of research.

Good Reporting of a Mixed Methods Study (GRAMMS) (O'Cathain et al, 2008)

- (1) Describe the justification for using a mixed methods approach to the research question
- (2) Describe the design in terms of the purpose, priority and sequence of methods
- (3) Describe each method in terms of sampling, data collection and analysis
- (4) Describe where integration has occurred, how it has occurred and who has participated in it
- (5) Describe any limitation of one method associated with the presence of the other method
- (6) Describe any insights gained from mixing or integrating methods

### 9.4.1 Describe the justification for using a mixed methods approach to the research question

A criticism made of mixed methods research has been that some studies fail to justify the use of a mixed method design (O'Cathain, 2008; Creswell et al, 2003). In order to address this Chapter 3 provided a methodological consideration of the different types of knowledge gained from using both qualitative and quantitative methods (section 3.2). Following this, a rationale and plan for using mixed methods was presented (section 3.3), and finally section 9.4.6 below considers the insights gained from using mixed methods. Therefore this thesis clearly describes (and evaluates) the justification for using mixed methods in order to address the research question.



interviews informed the development of mixed methods surveys, a critical evaluation of methods of analysis is provided in answer to questions in sections 9.4.4-9.4.6 which consider the integration of data.

The current research included interviews with and surveys of young people, parents and health professionals. These three participant groups were included with the aim of exploring the role of appearance from different viewpoints. Young people's own accounts are essential to understanding their individual experience of health and wellbeing (Rich & Ginsburg, 1999) and appearance research has highlighted the importance of subjective accounts (Rumsey & Harcourt, 2012; 2005). Parents of young people with NF1 were included in order to gain an understanding of the impact of NF1 on young people throughout their childhood and adolescence. Parents have been identified as highly significant within young people's management of chronic illness (Holmbeck, 2002), genetic conditions particularly (Williams, 1994) and appearance (Bellew, 2012). Health professionals were included in order to gain a perspective on their experience of working with a range of different young people and parents, as well as their knowledge of processes and procedures within health services. By integrating the findings from all three participant groups, a fuller and more detailed picture of experience was provided with both personal and professional accounts, these different perspectives illustrate one another. This section now considers issues that arose during the course of this research related to sampling and data collection, first in relation to health professionals and then young people and parents.

A particular consideration related to sampling was which health professionals to include in the research. As highlighted in the literature reviewed in Chapter 2, young people see many different types of health professionals in relation to their NF1. Initial discussions with members of support groups and clinicians who work with people with NF1 suggested that specialist nurses and genetic counsellors were a group with particular knowledge and insights into the psychosocial issues young people with NF1 manage. Therefore the exploratory interview stage of this research recruited genetic counsellors and nurse specialists. However, recruitment was challenging. There are a limited number of NF1 specialist nurses and genetic counsellors in the UK which meant that recruitment was taking place within narrow constraints. Furthermore, some concerns were raised (as discussed in Chapter 6) regarding anonymity and how easily identifiable comments may be. In addition several genetic counsellors responded with interest but were extremely busy and finding time for an interview was challenging.

After completing four interviews (three are included in the final analysis as discussed in Chapter 6) a decision was made that, while a greater number of participants may have initially been desired, themes were discernible across accounts. Essentially, the determination of sample size in qualitative research relates to the researcher's judgement and an evaluation of the quality of data in consideration of the use it is designed for (Sandelowski, 1995). Interviews were treated as in-depth expert accounts for the purpose of developing a questionnaire which would then survey a more diverse group of health professionals. The exploratory design of this research prioritises and builds on

individual in-depth personal experience and as such detailed contextualised expert interviews were appropriate within the research design.

When considering the second research stage, again which health professionals to include was a key consideration. While specialists may have greater insight into young people's experiences of NF1, the second survey stage aimed to generalise findings and as such a more diverse group of health professionals, who are representative of who young people see, was sought. This entailed reviewing literature, talking to clinicians and considering who young people and parents reported seeing in their interviews. It was noticeable that just a few of the young people interviewed were seen in specialist or genetics settings. However recruitment had not taken place through NHS clinics which may be relevant. A decision was made to circulate the health professionals' survey widely and include for instance orthopaedic departments where young people may be seen for scoliosis or pseudarthrosis as well as genetic counsellors and genetic departments. In addition a decision was made to recruit health professionals internationally.

A particular concern when planning this research was whether making surveys available internationally would mean that findings were less applicable due to the differences between healthcare systems. It is worth noting that there were challenges in analysing some of the data due to the international nature of the survey. For instance the roles of health professionals and how health systems work was clearly different and it was particularly difficult to quantify health professionals' roles and explore data on the basis of job type and nationality, particularly as 19% of respondents did not provide their nationality. However, this thesis concentrates on aspects related to the role of appearance and while there are certainly differences between healthcare systems the main focus was to gain an understanding of health professionals' perceptions of the impact of appearance related aspects of NF1 on young people. Health professionals' accounts of young people's concerns and management of NF1 were found to be similar across surveys when explored for differences based on nationality.

However, regardless of having opened up the survey internationally in order to recruit a more diverse group of health professionals, the analysis of surveys identified that the majority of respondents were genetic specialists. It is interesting to note that several (non specialist) health professionals who were contacted regarding the survey responded that, as they only saw a few patients with NF1 they felt they were unqualified to take part and suggested contacting specialist or genetics centres. Who cares for young people with NF1 and how qualified they feel in this role certainly appears to be an area that needs further consideration. While sampling had purposefully sought a diverse range of health professionals, it is also important to recognise that, in hindsight, a particular strength of this thesis may be that it provides an account of young people's experience from specialists. It is important to highlight that the views of health professionals are not being held up as the typical experience young people with NF1 may face but rather are included as the voice of expert health professionals in the area.

Recruiting young people to take part in research around appearance has previously been highlighted as challenging (Williamson et al, 2010; Wallace et al, 2007). In recognition of this young people and parents were recruited through a series of avenues. The first stage recruited through the general population in the UK while the second stage was international and included recruitment through NHS clinic lists (the rationale for this is discussed in Chapter 3). Although data saturation was deemed to have been reached (Guest et al, 2006; Parahoo, 1997) recruitment for interviews took a great deal longer than had originally been allocated and was more challenging than the survey study which exceeded the original numbers sought. This may relate, in part, to people preferring to take part in a survey as opposed to an interview, interviews demand more time and feel more involved. However, it is also likely to be related to the choices made regarding avenues of recruitment for each stage.

Recruiting in the UK for the interview stage, was in part due to a desire to conduct face-to-face interviews and use visual methods with young people. The rationale for using visual methods related to a desire to put participants at ease and address the power imbalances that can be present in a research interview, particularly for younger participants (Boyden & Ennew, 1997). However, as mentioned previously these methods were not found to be as engaging as hoped. It is difficult to be certain as to why this might be. One consideration was whether the age of participants impacted on their desire to use visual methods, as participants were older than originally planned; most were aged between 20-24 years. However, the use of timelines was not found to be related to age, one of the oldest participants used a timeline and one of the youngest did not. While some young people seemed reluctant to draw timelines, several brought photographs or photo albums to interviews and were keen to use images to share stories and explain different aspects of experience. Sharing photos was clearly important to some young people, many of the photos were of visible appearance changes and it may be that illustrating experience of appearance is better accomplished by literally illustrating accounts with photos rather than conceptualising experience within a timeline. This points to a need to explore new and innovative visual research techniques, including the use of photography, for engaging young people in appearance research (Harcourt, 2012).

The decision to recruit for interviews outside of NHS clinic lists was related to a desire to recruit a range of young people and parents. Individuals seen in specialist NHS clinics are likely to have more complex cases of NF1. This did lead to the inclusion of a range of young people and parents, some were seen by specialists and some were not. A few young people and parents were recruited through sites or message boards related to NF1, but not support groups, and had not had any contact with NF1 specialists or with support groups. As a researcher this held some challenges, in particular one participant had relatively little knowledge about NF1 and some of their comments about NF1 were factually incorrect. Ethically, a challenge was not correcting information; a decision was made to ensure that all young people and parents were given clear written information including sources of support at the conclusion of the interview. Recruiting young people through NHS clinic lists would have meant that participants could have been directed back to specialist professionals should such concerns arise. However, taking this approach may have lost some of the diversity of the sample.

Furthermore it is important to note that many young people with NF1 are not seen in specialist centres and therefore including these young people was a strength of the research.

The second survey stage did recruit young people and parents internationally and through three NHS clinics. One strategy that was particularly useful in promoting the survey was using facebook. A research page and profile was set up and, with permission of moderators, details of the survey were added to many NF1 facebook groups. Feedback suggested that participants liked being able to view the NF1 research page on facebook, and several commented that they liked to see the researcher's photo as it made it the process appear more personal. The survey was available online and those interested in taking part were able to click on a link from the advert directly to the survey. This meant that surveys were easy to access and may have encouraged participation. The majority of responses were online.

In hindsight, facebook could have been used earlier for interview recruitment, the page could have been set up and participants could have made contact through this platform. Another alternative to encourage participation might have been to consider email interviews (Gibson, 2010), although the reported high rates of learning difficulties in the NF1 population may prohibit this for some, so alternatives would have to be readily available. Another alternative would have been more telephone interviews. Two interviews were carried out in this manner, at the participants' request and appeared to work well. Research has suggested that telephone interviews provide similar depths of understanding as face to face interviews (Sturges & Hanrahan, 2004). Offering interviews by telephone or email (or both) may have increased the speed of recruitment. However, it is important to note that interviews in the current research were of a sensitive nature and in several interviews participants (both parents and young people) became visibly upset. In a face to face interview context cues can be read and previous experience drawn upon to handle such situations sensitively, whereas in an online or telephone interview, nuances can be harder to pick up on. In one telephone interview in the current research, a participant became upset and began crying. In a face to face interview it would have been clearer that the participant was becoming upset but over the telephone this was harder to read, and until the participant was audibly crying, it was not clear that they were upset. However, if individuals are concerned about their appearance then the option of an interview that is not face to face may be preferable. Furthermore, it is important to note however that while recruitment took longer than initially planned, the final numbers of young people interviewed are comparable to similar studies (for instance Wallace et al, 2007).

Another sampling issue related to the inclusion of young people under 16 years in interviews and under 18 years in surveys. In the current research a number of young people under the specified ages indicated they wished to take part in the research but parental consent was not provided, it was unclear exactly why these parents did not wish their child to take part. Previous appearance research with young people and parents has also reported challenges involved in recruiting young people and points to parents concerns about their child's participation (Williamson et al; 2010). During the course of this research a number of patient days and support group meetings for people with NF1 were

attended by the researcher in order to disseminate research findings. On these occasions the researcher took the opportunity to informally ask parents with younger children with NF1 about research participation. Parents suggested that they would be concerned about their children learning information about their condition that they were not ready for and future research may wish to consider addressing this. This could be achieved by explicitly writing on information sheets that interviews would be participant led and no new areas of information would be introduced.

Another point to note is that during dissemination of this research a few health professionals have suggested that the young people in this research are a well educated group of individuals compared to those that attend clinics. However, it is difficult to suggest who a typical NF1 patient is, due to the variability in the condition. Individuals attending NF1 specialist clinics in the UK are likely to be those with the most complex needs and while many of the young people in this research reported seeing specialists, many did not. This may be a particular strength of this research, making the young people more representative of young people with NF1. However it is difficult to be certain. It is important however to recognise that participation in research into sensitive topics such as this, is likely to have a particular bias. Individuals who are finding a situation challenging may be less likely to participate in research but, if research is to be applicable it must strive to include the views of those affected (Alexander, 2010). Certainly many of the young people and parents interviewed were highly knowledgeable about their condition and were engaged with support groups, but this was not the case across the board.

On several occasions both young people and parents in the same family were interviewed. Some young people chose to have a parent stay with them during their interviews, so it is therefore difficult to be certain if or how this might have impacted on findings. Interviews were examined particularly looking for differences between interviews with and without parents present. There were no obvious differences; however this does not mean that parents being present did not influence interviews. On a few occasions parents prompted young people towards answers which was often supportive and helpful of the interview process. However this did mean that answers were potentially different to those the young person may have given without the parent present. This was more apparent at the start of interviews as rapport was being built between the interviewer and the young person and lessened during interviews. The researcher has a great deal of experience working with children, young people and parents and various techniques were employed in order to ensure that the young person's view was being represented.

A final point to reflect on regarding sampling is the impact of not having asked whether young people had ASD/ADHD or associated learning difficulties. This was a difficult decision that was revisited throughout the research. While previous research clearly highlights the importance of ASD/ADHD and associated learning difficulties within young people's experiences of NF1 (Pride et al, 2012; Barton & North, 2004) it also suggests this impact is complex (Isenberg et al, 2013; Garg et al, 2012). Simply categorising young people by diagnosed learning and behavioural difficulties was deemed to be too simplistic. Interview findings highlighted the diverse ways in which learning difficulties may impact on experience but also highlighted the difficulties inherent in categorisation. Several young people were

diagnosed late (one in their 20's) while one parent explained that her child did not have an official diagnosis but was treated by school as though she had ASD as that was how she presented. Some participants (parents and young people) felt an official diagnosis and label was useful in gaining support, others explained they did not want to be categorised and had chosen not to explore that area. A decision was made that learning and behavioural difficulties would not be explored in the survey, this is not to suggest that learning and behavioural difficulties are not highly relevant for many young people. Particularly in light of findings presented in this thesis regarding the importance of social skills and management of social interactions, the prevalence and impact of learning and behavioural difficulties is an area in need of further research and definition. However, an exploration of this aspect is beyond the remit of the current research.

With regards to analysis it is important to note that there are limitations inherent in the use of multiple statistical tests. Interpreting the probability of multiple tests is difficult; if the probability threshold is 0.05 for each comparison then multiple tests increase the chance of obtaining statistically significant results even if the null hypothesis is true. If 13 independent comparisons are performed the chances of obtaining at least one significant result, simply by chance, is approximately 50%. In order to address this one approach is to set a stricter threshold for statistical significance. In the multiple t-tests reported in chapter 8 results were not significant and as such this would not have altered the findings. Another way to address the challenges of multiple tests is to ensure that only planned comparisons are carried out. In the current research all analysis was guided explicitly by the research questions formulated in Chapter 7. By focusing in this way on making scientifically sensible comparisons rather than exploring every possible comparison the statistical power of comparisons is increased (Keppel & Wickens 2004).

In conclusion, while there were challenges involved in sampling, data collection and analysis the strengths of this research lie in the use of different methods that have explored the views of health professionals, parents and young people. Participants are geographically diverse and seen in different settings by different health teams. Surveys were recruited through clinics in the UK and internationally through professional organisations, support groups and using online communities.

#### **9.4.4 Describe where integration has occurred, how it has occurred and who has participated in it**

Specifically addressing how methods are integrated within a mixed methods research project has been described as crucial (Bryman, 2008; Creswell 2003). A criticism of studies has been that little attention is paid to how integration of data occurs (O'Cathain, 2008). In order to address this, Chapter 3 described clearly how methods of investigation would be used and integrated in this programme of research and then the process of integration was described clearly as it occurred. Finally, this section reflects on how integration took place within this research and the following sections (9.4.5 & 9.4.6) consider the challenges and benefits of this approach. Methods were integrated at three points which are considered below:

- (1) Themes from qualitative analysis were used to identify questions and variables for surveys in Chapter 7

The first point of integration involved several steps that are outlined in chapters 4-7. The first step was to identify patterns across each interview study which could be explored further within a questionnaire. This entailed thematically analysing each interview study; this method was well suited to this task due to its theoretical flexibility and its ability to systematically organise individual experiences into patterns (Baraun & Clarke, 2006; Boyatzis, 1998). In this thesis the thematic analysis took an inductive approach to analysing data, themes are presented and discussed in chapters 4, 5 and 6. Having analysed each data set the next step was to consider themes concentrating specifically on aspects related to appearance. In practice this meant reviewing data and themes with appearance as a filter. This filtering process is presented within the discussion section of chapters 4-6 and led to the identification of the key aspects of each groups experience which are presented at the end of each interview chapter (figures 5, 7, 9) These key aspects were then used to develop a series of research questions presented in Chapter 7. The next step was then to consider how research questions could be explored further within a survey and identify areas of literature that mapped onto these aspects. This then led to the identification of specific measures, quantifiable questions and open ended questions. The process as a whole is documented in appendix 17, 19 and 21.

- (2) The reporting of survey findings in answer to the research questions in Chapter 7 contained both qualitative and quantitative findings

The use of mixed methods in the surveys enabled quantification of earlier findings as well as allowing for new findings. Findings were integrated and reported in answer to specific research questions. For instance the research question '*How do young people with NF1 feel about their appearance in general and do they report their condition as noticeable?*' was answered by using a standardised measure of body esteem which was analysed statistically, a quantifiable yes or no question regarding noticeability as well as open ended questions about young people's concerns and what NF1 meant for them which were analysed using content analysis. By presenting data in this way, in answer to specific questions, the applied and pragmatic approach underlying this research is highlighted.

- (3) The discussion of findings and application of findings in this current chapter has integrated both qualitative and quantitative findings

The current chapter discusses findings from across this thesis considering knowledge gained from quantitative and qualitative methods in order to consider the impact of appearance on young people with NF1 and how this knowledge could be applied. Section 9.4.6 reflects on the knowledge gained from integrating findings in this manner.

At each of these three points how data is integrated is made explicit. Dures et al (2010) have stated that there is a lack of guidance and advice for researchers regarding the mechanics of mixing methods. This thesis has sought to address this by clearly describing the aims behind using mixed methods, describing in detail how methods were mixed and then reflecting on and evaluating the processes involved. The researcher carried out all points of integration within a supervisory team. All interpretations were shared and discussed in order to ensure rigour of analysis. Having reflected on how methods were integrated in this section, the limitations of integration are considered below and the utility of a mixed methods approach overall is considered in section 9.4.6.

#### **9.4.5 Describe any limitation of one method associated with the presence of the other method**

Qualitative and quantitative methods were used both sequentially (interviews to surveys) and concurrently (in the surveys). Overall, the methods integrated well and the presence of each method supported and enhanced the other (see section 9.4.6 below). It was helpful to have decided, prior to commencing research, the priority and role of methods, in particular, the role of less dominant methods. For instance, open ended questions in surveys were included specifically to illustrate certain points and allow for new findings; the purpose of these questions was defined clearly in advance (see section 7.3). Open ended questions in surveys have been found to produce a more diverse set of answers than closed questions (Reja et al, 2003) and without a clear definition of the purpose of findings open ended questions can be difficult to code and interpret (Sudman & Bradburn, 1991). Defining the role of questions meant that findings were used for specific purposes reducing the limitations that can be associated with open ended questions within a quantitative survey. Without this clear definition open ended questions may have been difficult to analyse and may have had little impact within a primarily quantitative survey.

There were challenges in using one method to inform another, specifically in terms of mapping some quantitative measurements on to qualitative phenomena. One particular difficulty related to measuring noticeability. In line with previous research (including Moss, 2005) interviews appeared to suggest that appearance related concerns did not always relate to objective assessments of young people's appearance. Young people themselves described how smaller, non visible differences could be just as challenging, or more challenging, than noticeable differences. However parents reported that the extent of noticeability was important to their child. Perceived noticeability was therefore taken forward and measured quantitatively in surveys. Reflecting the different ways young people and parents discussed the concept young people were asked if they felt their condition was noticeable with a response choice of 'yes' or 'no' while parents were asked to rate how noticeable their child's NF1 was to others on a scale of zero to ten. This meant that findings were analysed using different statistical tests. A yes/no question on the young peoples' survey meant that noticeability was investigated using t-tests while the scale used in the parents' survey meant that findings were investigated using correlations. This presented some difficulties when then comparing the survey findings. In hindsight the surveys could have asked young people and parents the same question and measured responses using the same format. The desire to be grounded in the detail of personal individual in-depth data

can conflict with the desire to produce clear and robust statistical analysis. In this case rather than reflecting the different ways participant groups discussed noticeability measurement should have taken place through a consideration of how it would be compared.

A second challenge related to mapping psychological constructs onto qualitative findings. The importance of reconceptualising NF1 through adolescence was defined as a key aspect of young people's experience in Chapter 4. This was taken forward quantitatively using the B-IPQ in order to explore young people's (and parents') perceptions of NF1. In contrast to the personal descriptions of the young people interviewed, illness perceptions were not found to change with age in the analysis of survey data. However, they were found to be highly varied and this may be an important factor in interpreting findings. The detail of individual changes in perceptions described in interviews may not be accounted for in exploring correlations between young people's perceptions and age. The reconceptualising described in interviews may be better measured longitudinally by exploring how individual perceptions change overtime, rather than exploring if perceptions correlate with the age of participants. How individuals understand and cope with NF1 is likely to be highly individual and as such future research should consider ways of quantitatively exploring individual changing perceptions rather than if there are differences by age.

#### **9.4.6 Describe any insights gained from mixing or integrating methods**

This programme of research has benefited greatly from employing both qualitative and quantitative methods. The different methods have produced different types of knowledge, in answer to specific research questions, providing an understanding of the role of appearance within young people's experiences of NF1. This is an area of research that has not been explored previously. A survey based on the existing literature regarding young people's experiences of NF1 would have been challenging, due to the lack of research with this age group. Furthermore the focus of this particular programme of research was specifically on the role of appearance. The aim of starting with in-depth interviews was to explore the ways in which appearance played a role across experience, being open to new findings. This research is therefore grounded in qualitative data and can be characterised as qualitatively driven (Mason, 2006). In order to evaluate the qualitative underpinnings of this research, Yardley (2000) suggests a series of characteristics of good qualitative research. These are considered in terms of the current research in table 25.

##### **Sensitivity to context**

- Interview guides were developed following a review of literature related to appearance, NF1, research with young people and health and illness during adolescence.
- Health professionals working in the area, including the board of The Neuro Foundation, were supportive of the research.
- Consideration was given to how findings from health professionals' interviews would be reported so as not to identify individuals.

<ul style="list-style-type: none"> <li>• Participatory methods were offered to young people, in order to engage them and address power imbalances inherent in researching young people's experiences</li> <li>• Ethical concerns were given a great deal of consideration and are addressed in each study</li> <li>• Sensitivity to the potentially emotive issues were considered and guides were broad in nature</li> <li>• Some aspects of experience were not addressed with young people (eg inheritance) so as not to introduce areas of concern</li> </ul>
<p><b>Commitment and Rigour</b></p> <ul style="list-style-type: none"> <li>• The researcher has developed relationships with support groups, other researchers in the field and with specialist health professionals in order to engage with the subject area.</li> <li>• NF1 patient events have been attended, in some cases to disseminate findings and at other times to engage with the community.</li> <li>• The researcher has a great deal of experience working with children and young people</li> <li>• Training in specific skills (including visual methods) was undertaken prior to research commencing</li> <li>• Different participant groups and methods of investigation demonstrate the breadth and depth of analysis undertaken in this programme of research.</li> </ul>
<p><b>Transparency and coherence</b></p> <ul style="list-style-type: none"> <li>• The use of methods is fully considered in Chapter 3 and evaluated in a discussion of the interview studies the end of chapter 7 and in the current chapter</li> <li>• How interviews were undertaken is clearly described in each individual chapter</li> <li>• How methods have informed the development of surveys is clearly described (section 7.3)</li> <li>• Analysis contains direct quotes from interviews in order to evidence findings</li> </ul>
<p><b>Impact and importance</b></p> <ul style="list-style-type: none"> <li>• This research was undertaken following a review of all the current literature and addresses an area identified as in need of further research by those working in the area</li> <li>• Findings are discussed in relation to how they were used in a survey (Chapter 7)</li> <li>• Findings are integrated with quantitative findings to provide a theoretical discussion of findings overall</li> <li>• Findings are integrated with quantitative findings to provide a consideration of the practical application of findings overall</li> <li>• Findings have been disseminated in a written paper published in 2013 (appendix 29) and through presentations at conferences and patient and support group events (appendix 30)</li> </ul>

Table 25: An evaluation of the quality of the qualitative components of the research Using Yardley's (2000) Criteria for Assessment of Qualitative Research.

The characteristics outlined by Yardley and evidenced in table 25 underpin the research programme overall and demonstrate how by starting from an in-depth qualitative viewpoint, sensitivity to context; commitment and rigour; transparency and coherence; impact and importance are integral to the research overall. In-depth participant led interviews identified many rich and varied accounts of young people's experiences, from three perspectives. Thematic analysis determined areas of commonality across each data set, this method of analysis allowed rich individual in-depth data to be categorised into units of meaning (Braun & Clarke, 2006). This provided a framework for the development of

surveys (Creswell, 2003). Using thematic analysis in this way has meant that the aspects measured in surveys are grounded in the personal detailed experience found in interviews.

Validated measures were used within parents' and young people's surveys and data was analysed using well known statistical tests and software. Quantitative findings are reported in line with widely used recommendations and conventions (Pallant, 2010). Analysing surveys statistically has provided valid and reliable and replicable findings. Statistical analysis has been able to explore relationships both within individual surveys and between surveys. Furthermore, the qualitative analysis of open ended questions provided space for new observations and explored questions raised in the interview stage further. This supported the presentation of findings from surveys. Research questions were answered with both qualitative and quantitative findings; however, it is important to note that the survey was primarily quantitative, at this juncture quantitative methods were more dominant.

Discussing the role of appearance within young people's experiences of NF1 and the application of findings has benefited greatly from combining both the qualitative and quantitative stages. The two data sets illustrate one another and deepen overall understanding. For instance, in discussing body esteem and appearance evaluations, quantitative findings demonstrated body esteem to be important within young people's experiences but not highly negative. By exploring qualitative data this finding was explained in terms of appearance concern relating to uncertainty and future changes. This was then tied in with quantitative findings regarding the low levels of control expressed on the B-IPQ. By combining the two stages of research a fuller picture is gained than either of the two individual stages could have provided singularly. Furthermore, by also being able to consider findings from parents and health professionals, layers of explanation are added, deepening the understanding and adding depth and definition.

Using mixed methods in the survey gave space for new findings at this stage. For instance almost a quarter of the young people who responded reported that their main concern related to having children (n=15, 23%). The possibility of having children with NF1 had been discussed by a few participants in earlier interviews but the finding that such a large number felt this was their main concern was surprising, particularly as just one health professional reported this as young people's greatest concern. However, it is important to note that inheritance was not a focus of this research and furthermore this was specifically not explored in interviews due to ethical considerations. Yet this does highlight that reproductive concerns may be important to young people, and this would not have been found if the survey had not included open ended questions. It is unclear from the current research if and how these concerns impact on young people but it would appear to be in need of consideration in future research. The importance of inheritance was generally an interesting difference between health professionals and both parents and young people. While health professionals felt that this impacted on families' management and experience and whether appearance was a concern, in both interviews and surveys, using statistical analysis of parents' and young people's findings, this was not found within parents' and young people's survey findings.

However, it is acknowledged that this needs greater consideration and in the current research programme questions related to inheritance may have been interpreted in different ways by different participant groups. Furthermore, health professionals reported that differences were particularly apparent at initial diagnosis and survey respondents (both parents and young people) were unlikely to have been recently diagnosed.

The use of mixed methods enabled the qualification of the impact of specific key aspects of living with NF1 which it is hoped will be useful to those who work with young people with NF1. While individual in-depth accounts are highly important in understanding individual lived experience, being able to quantify this and understand how common or uncommon such experiences are, is necessary in order to plan and care for individuals within health care and for support groups.

#### **9.4.7 Section Conclusion**

Adopting a mixed methods approach and including different participant groups has benefited the research findings in four primary ways. First, aspects identified in in-depth qualitative interviews were explored through statistical analysis of survey findings meaning that the research is rooted in individual experience and quantified statistically. Second, findings between different participant groups have strengthened one another (such as the importance of social interactions) or have pointed to areas that may need further exploration (for instance the different perceptions of the role of noticeability). Third, continuing the use of mixed methods in the survey provided new findings (for instance reproduction being a concern for many young people) and finally, findings across the different stages of research have been combined to create a detailed picture with different aspects of the data illustrating one another (such as the importance of appearance).

#### **9.5 Areas of further research**

This research has provided an in-depth understanding of how issues related to appearance impact on young people with NF1, it has also raised a plethora of questions that need further consideration. Whilst areas have been suggested throughout this chapter, this section identifies and discusses three key areas in need of further consideration and asks how they might be explored. First, greater understanding of the way in which young people and parents manage uncertainty and noticeability is proposed, second it is suggested that there is a need to obtain a detailed picture of which health professionals are involved in the care of young people with NF1. Finally the value of including participants as advisors in future research is considered.

##### **9.5.1 Managing uncertainty and the importance of noticeability**

Managing uncertainty and feeling a lack of control over NF1, particularly related to appearance changes, were evidently important to many young people and parents. Further understanding of how individuals manage the possibility of an altered appearance and uncertainty during adolescence is

needed in order to develop interventions to support resilience and plan how and when these should be delivered.

Frith et al (2007) discuss 'anticipatory coping' (preparation for managing an event that has not yet occurred but may do so) as a mechanism used by participants in their study with patients who were expecting to lose their hair whilst undergoing chemotherapy. A first stage in anticipatory coping has been identified by Aspinwall and Taylor (1997) as proactive coping which involves accumulating resources (including information) and skills to prepare in general rather than manage a specific stressor. This is an active process and informational support is identified as particularly beneficial. In discussing the application of findings, making information available for young people and parents in relation to the appearance related aspects of NF1 was highlighted as an important area to consider. An evaluation of the informational needs of young people and parents through adolescence may therefore be a particularly useful first step in developing any such resource and planning delivery. This could involve first asking support groups and health professionals what information they have available for young people. Focus groups of young people and parents (in separate groups) could then be asked to examine and discuss this information provision as well as exploring more broadly how and when they would have liked information to have been provided. Such focus groups could be facilitated online in order to encourage engagement. Specifically, any such evaluation should consider what information is needed, when it should be delivered, how it should be delivered and by whom.

In addition to exploring the value and use of practical information it is also important to consider that the meaning of an altered appearance and perceptions of a medical condition are likely to alter during adolescence. The development of abstract thought during this life stage may alter the way in which young people perceive the implications of NF1 (Sebold et al, 2004). During interviews some young people described adolescence as a time of reconceptualising their condition. For some this led to an increased awareness of the appearance-related aspects of the condition and anxiety about whether changes would occur. However differences in perceptions by age were not found in the survey of young people. A possible explanation for this is outlined in the previous section which suggested that due to the diversity of illness perceptions evidenced in the survey individual experience across adolescence should be explored. This could be achieved using qualitative methodology and interviewing young people regularly during adolescence in order to explore individual lived experience through this life stage. Longitudinal qualitative interviews are important in understanding how people experience, manage and respond to change (Hermanowicz, 2013). They have been used previously with children, young people and adults, and health is described as an area particularly well suited to the employment of this method (Holland et al, 2006). However, such methods are very time consuming and particularly when considering such research with young people ethical considerations, including gaining and maintaining consent, would need attention (Warin, 2011).

Findings presented suggest that the meaning and impact of the noticeability of NF1 is an area in particular need of further exploration. There were no differences in young people's accounts of body esteem, social comfort and stigma according to whether they reported their NF1 as noticeable to

others or not. But parents' ratings of noticeability correlated with their accounts of their child's stigma experiences and social comfort. A possible explanation for this may be that parents and young people are considering the impact of noticeability on social interactions in different ways. Parents may be considering how others react to their child and noticeability is likely to impact on these situations. A more noticeable difference may lead to greater reactions from strangers. For young people the impact of noticeability on social interactions was described in terms of worrying about how others might react rather than managing actual reactions. In these circumstances objective noticeability would not necessarily impact. However, as discussed in section 9.4.5 the different ways in which noticeability has been measured in surveys makes it difficult to explore this further. Future research should look to investigate what young people and parents mean by 'noticeable' before constructing a way to measure subjective noticeability. It would be particularly useful, if planning to compare accounts, to measure in a similar way using a robust measure.

Such a measure should then be used to explore how and why perceived noticeability becomes important to parents and how this might impact on the salience of appearance for young people. It has been suggested in this thesis that the importance of noticeability for parents may be related to monitoring their child's condition for appearance changes which may, in turn, lead to an increased focus on appearance. Moss and Rosser (2012) highlight how cognitive biases in processing appearance evaluations can lead to an increased awareness of appearance related information. However whether such processing biases exist for parents of young people with a visible difference has not been explored. If such a bias exists this may lead to an increased emphasis on appearance for parents, which may impact on young people's own appearance evaluations. The importance of parents in the formation of young people's body image is well documented; influences can be internalised by young people and become part of their body image (Gillen & Lefkowitz, 2009; McCabe & Ricciardelli, 2001). Even if young people do not internalise a parent's awareness of noticeability an increased focus on appearance may result in poor adjustment (Lawrence et al 2006a; Moss & Carr, 2004).

Longitudinal research with families should therefore explore the meaning of appearance and noticeability through both interviews and validated measures during childhood and adolescence. Within this research should specifically consider if and how parents' ratings of noticeability influence how young people feel about their appearance. Research should explore if there is a relationship between parents accounts of noticeability, the importance placed on appearance by both parents and young people and general wellbeing. A particularly interesting development to use within this may be the NF1 specific quality of life measures for parents and young people which are reported as under development (Nutakki et al, 2013). This may be beneficial in exploring how NF1 impacts longitudinally in terms of what specific domains NF1 impacts on and how noticeability, individual perceptions, appearance and social interactions correlate with such a measure.

### 9.5.2 Health professionals

Current UK guidance (Ferner et al, 2007) is that children diagnosed with NF1 should have regular health checks with a paediatrician throughout childhood. Adult care is then transferred either to a GP, a specialist NF1 clinic or another team depending on individual need. Young people are therefore likely to be seen in many different settings by many different specialists, particularly after they have transitioned to adult health care. A challenge in disseminating information and applying findings from this thesis may therefore lay in identifying who the health professionals that work with young people with NF1 are, and then in reaching such a diverse group of professionals. Many of the professionals working with young people with NF1, such as GPs, may only care for a very few individuals with the condition during their career, and as such having detailed knowledge of all aspects of every condition is not feasible. Furthermore while some health professionals may be aware of the psychosocial impact of appearance related concerns for their patients, Rumsey (2008) suggests that they may be uncertain about how to support these concerns.

This suggests a need to obtain a clear picture of the views of health professionals who work with young people with NF1. This would involve large scale surveys of the different health settings in which young people might be seen. In the survey study, aside from NF1 specialists, young people and parents reported being seen by professionals including general practitioners, genetics services, eye specialists, paediatricians, dermatologists, neurologists, pain specialists and orthopaedic specialists. These professionals could be targeted and sent a very short online survey asking for their area of speciality, whether they see young people with NF1, what ages they see and roughly how many and how often. This survey could be sent through professional organisations including genetics organisations and dermatology bodies. In addition to identifying how to target support and training this information could also allow for future research exploring the views of non NF1-specialists. As noted previously, while the health professionals in the current research appeared to be aware of the issues and concerns of young people and parents, respondents were mainly specialists and this did not reflect who young people and parents reported seeing. However, while many young people with NF1 are cared for by non-specialists, it is interesting to note that several of the non-specialists approached in this research reported that they were not the right people to answer questions about NF1, as they only saw a few individuals with the condition.

This research has suggested that genetic counselling in particular is an area in which young people could gain support in adapting to NF1 and the possibility, or actuality, of an altered appearance. However, there is limited research that has explored young people's experiences of genetic counselling. There is a growing body of research that has started to explore young people's experiences of genetic diagnosis (Callard et al, 2012; Duncan et al, 2007; 2008; Gaff et al, 2006), however young people's experiences of genetic counselling for an ongoing condition is not clear. Any research in this area should explore if and how young people are offered and take up genetic counselling as well as asking if this is the right medium in which to provide support. In addition

research should consider how and when referrals to genetic counseling should take place. Any such research should include genetic counsellors themselves and should consider the cost implications of such a service. Research in this area would benefit from being country specific in order to explore the specifics of the health services of an individual country.

Once research has identified which health professionals work with young people with NF1, their training needs and the accessibility of additional supportive services, including psychological services should be audited. In line with findings presented by Williamson (2012) in her research with health professionals working with young people with appearance changes resulting from cancer, participants in the current research pointed to inconsistencies and difficulties in accessing support services. Williamson suggests a need for clear appearance care pathways within the NHS, as well as training for health professionals and a directory of appearance related specialist services. Parents in the current research reported that access to a trained counselor would be greatly appreciated, yet health professionals reported unclear pathways and difficulties accessing psychological services. Further research is needed to explore whether the current provision meets the needs of young people, parents and health professionals. Specifically, research should explore if and how appearance concerns are identified and managed. This could involve telephone or emailed surveys of health professionals who work with young people with NF1 asking if their patients express any appearance anxiety and if they do where do they refer them for support. The services suggested by health professionals could then be approached and asked about the availability of support and resources available for young people.

### **9.5.3 Participant and public involvement**

An important aspect of this research has been the researcher's engagement with individuals with NF1, health professionals who work in the area, parents groups and support groups. Building relationships within the NF1 community has been an essential part of this PhD. This has enhanced the research and provided the opportunity to discuss ideas and plans with young people, parents, health professionals and others who were interested. Listening to and engaging with those who are involved in research in a meaningful way is likely to enhance the quality of appearance research (Bates, 2012). At different stages of this research representatives of participant groups were consulted. For instance meetings with health professionals took place prior to commencing research and throughout the research process. Young people and parents who participated in interviews were asked for feedback on questionnaire development. Patient events, conferences and support group events were attended by the researcher, partly to disseminate research, and this also provided an informal opportunity to talk to all participant groups about the research and plans. This supported the research particularly in terms of identifying how and where to recruit people to take part in the research. Several individuals chose to promote the survey through social media and share information with groups. This was done without being asked but rather is a natural way to engage with social media. Formalising this type of patient and public involvement activity and engaging with participants from the early stages of planning may have supported recruitment of the interview stage.

This could also support the design of research methods, for instance discussions with young people in the early stages of this research may have led to a change in the visual methods used in interviews. Furthermore, discussions with parents at patient days identified concerns regarding their children's participation in interviews, had such conversations occurred earlier on in the process changes could have been made to information sheets.

The consultations with participant groups in the current research were informal, future research should consider making these consultations more formal and embedding them within the research design. NICE guidelines (2013) highlight the value of including participants' views in the development of research. Participation needs to be meaningful and for research to engage with populations, such as those in this research who can be difficult to reach, the views of those who researchers are looking to engage with should be sought. However, participation should be considered carefully so that all involved are clear regarding their roles. Furthermore, it is important to note that engaging people in patient and public involvement in research can be challenging. Challenges may include negotiating practical considerations such as reimbursement of service users time and travel costs and ensuring that those individuals who engage in participant and public involvement are reflective of the population that is being researched and have the appropriate skills to engage in research (Staniszewska & Denegri, 2013). Engaging with participant groups is also time consuming and as such needs to be clearly identified and assigned time within the project. However while there are challenges, there is support and advice available for researchers looking to include public involvement in their research. There are 'how to' guides and advice available online for researchers considering engaging in these ways (including the Research Design Service North West online guidance: [www.rds-nw.nihr.ac.uk/publicinvolvement/how-to-guide](http://www.rds-nw.nihr.ac.uk/publicinvolvement/how-to-guide)) and organisations such as INVOLVE ([www.invo.org.uk](http://www.invo.org.uk)) provide a range of publications, advice and support.

Future research should actively involve young people, parents and specialist health professionals from the planning stage onwards in order to support the practical development of research and subsequent development of interventions and guidelines. In addition to advising regarding practical considerations, involvement of this type makes research more robust and credible (INVOLVE, 2013).

## **9.6 Conclusion**

This research has demonstrated the value of including different participant groups and using a mixture of research methods in order to obtain a detailed understanding of the role of appearance within a highly unpredictable and varied condition that can or does alter appearance. The pragmatic approach taken is particularly valuable in obtaining findings that reflect individual experience but are also applied and offer potential benefits to patients in the future..

This thesis has made an original contribution to knowledge regarding of the role of appearance within young people's experiences of NF1. This programme of research has demonstrated that how young people with NF1 feel about their appearance and how aspects of appearance impact on social

interactions are highly relevant to their wellbeing. Yet while appearance evaluations are important, they are not always negative; concerns relate to future appearance changes rather than noticeability. Adolescence is described as a time of re-conceptualising NF1 and this may provide an opportunity in which to address appearance concerns and support resilience; both young people and parents report a desire for information and support at this time. Supportive care and information should particularly address uncertainty about future appearance changes, and support young people to manage social interactions, particularly other people's reactions and talking about the appearance related aspects of NF1. Findings highlight the important role that specialists play within young people's (and parent's) adaptation to and understanding of NF1. Findings suggest a need to further explore uncertainty and control within the management of conditions that could alter the appearance and point to a need to further investigate the different importance placed on noticeability of visible difference by parents and young people with conditions that could or do alter appearance.

This thesis contributes to the literature by providing data about a group who are under researched about an aspect of their experience that has not been explored. A detailed understanding of the role that appearance plays within young people's experiences of NF1 has been provided and the potential for improving the provision of supportive care for this group and the need for further research has been evidenced.

### **9.7 A final personal reflection**

In discussing the approach of this thesis I commented on how researchers own lives and experiences shape their research. It appears to follow therefore that the research we do will also impact on our personal biographies. This brief section considers how this research has impacted on me on a personal level.

Throughout his research I have met many people (parents, children, young people, health professionals and researchers) who have made me step back and really think about the choices I have made and how I have responded to events in my own life. Meeting parents of young people in their 20's and hearing their experiences has sometimes made me worry about parenting a young person with a chronic condition and at other times I have felt reassured and my opinions and choices have been validated. Hearing the stories of young people with NF1 was sometimes difficult. My background is in listening to young people in order to support and help them, in a research context just listening without trying to problem solve was particularly challenging for me.

At times during this research I felt I identified strongly with many of the emotions expressed by participants; both parents and young people. I felt particularly emotional when parents described their experiences of their child's diagnosis and coming to understand what it would mean. This has made me very aware of the fact that I have never really spoken about when my son was very ill and first diagnosed and I admired the parents who spoke to me so openly about such private experience. I also realised that I have not met many people who have been through similar experiences. There were a few occasions where I had to stop myself from nodding and agreeing with parents. I have

reported (in chapter 5) that some parents described how small incidents could take them back and they would express strong emotions that they had felt during the period of diagnosis. I found this to be true for myself, several interviews left me remembering and reflecting on some of the very difficult and challenging moments I had when my son was in hospital and during the early years of diagnosis. It made me reflect on how lucky I am that things are not as scary as they were at that time but also made me a little scared that they could be again. I also found myself sometimes mentally noting what young people felt had been helpful or unhelpful and thinking about my own parenting style.

Overall I hope that my personal enthusiasm and engagement in this research has been beneficial to the research process, I know that doing this research has made a great impact on me, as a researcher, a parent and as an individual.

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**Appendix 1: General recruitment advert for interviews with parents  
and young people**

**Would you be willing to take part in a research interview?**

The Centre for Appearance Research at the University of the West of England is researching young people's experience of living with Neurofibromatosis type 1; it is expected that this work will be used to help develop support for people with NF1 in the future.

We would like to speak to people (and the parents of people) aged 14-24 who have NF1. All interviews will be anonymous.

**To take part** in this research or to receive **further information** please call **Jenny** on 07500 121137

or

email [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk)

**THANK YOU**

## **Appendix 2: Recruitment information from The Neuro Foundation Newsletter regarding interviews with parents and young people**

Would you be willing to take part in a research interview?

The Centre for Appearance Research at the University of the West of England is Researching young people's experiences of living with Neurofibromatosis Type 1; it is expected that this work will be used to help develop support for people with NF1 in the future.

We would like to speak to people (and the parents of people) aged 14-24 who have NF1. All interviews will be anonymous.

To take part in this research or to receive further information please contact:

## **Appendix 3: Information sheets for young people (interview study)**

### **Young Peoples' Experiences of Neurofibromatosis Type 1: An Exploration of Psychosocial Issues**

#### **Information Sheet for young people**

You are invited to take part in a research study. Before you decide whether or not you would like to take part it is important that you know why the research is being carried out and what you will be asked to do. This sheet will give you the information you need so you can decide if this study is something you would like to take part in. If there are any points you are not clear on please ask. Please read the sheet carefully, you may want to discuss this research with your family; if you are under 18 please share this information with your parents/carers as we will require their consent for you to take part. Thank you for your time reading this sheet.

#### **What is the purpose of the study?**

Neurofibromatosis type 1 (NF1) is very variable. Some people may not even know that they have NF1. For other people some aspects such as the changes to appearance that can occur may cause more difficulty. We would like to understand how young people experience having NF1. There has not until now been much research that focuses on how NF1 impacts specifically on young people. If we can understand how young people with NF1 feel and how they manage their condition then we can try to help to make things better for other young people and their families in the future.

This study is being carried out by researchers at the Centre for Appearance Research at the University of the West of England. This study will be part of a research degree (PhD) for one of the researchers.

#### **Who can take part in the study and do I have to?**

You are being asked if you would like to take part as you contacted the research team for more information after seeing one of our adverts. We are interested in talking to young people aged 14-24 who have a diagnosis of NF1.

You do not have to take part. It is totally up to you and if you are under 18 we would also need your parent/carers permission. If you do decide to take part you will be asked to read and sign a consent form saying you would like to take part. If you change your mind about taking part you can stop at any time; no one will mind or be upset.

#### **What will I have to do?**

If you decide you would like to take part we would like you to meet with a researcher at your home or somewhere else where you feel comfortable talking at a time that's good for you. It won't be a formal interview and there are no right or wrong answers. The researcher will ask you some general questions about yourself and will ask you to make a timeline with them. This would be working together with a piece of A3 paper thinking about the events that have happened in your life in general and related to NF1 that you think are important. You could keep the timeline if you like although the researcher would like to take a picture with your permission. (For more details see the sheet "what is a timeline?")

We understand that some of this topic may be personal and sensitive and hard to talk about, it's fine if you don't want to talk about certain things and if you feel uncomfortable you can stop at any time.

We would like to tape the conversations; these will be written up and then destroyed when the study is finished. Only the researchers on this study will be able to listen to the tape and your name won't be linked to it. If you'd rather the researcher could take notes on the conversation instead.

**What are the benefits?**

We don't expect there will be any direct benefits for you; some people find it interesting to take part and to talk about their experiences. The information will help us to understand your experiences and write a questionnaire that will be used in further research. We would like to make sure we're asking the right questions about NF1 and we hope that overall this research programme will help to inform the provision of care and support for young people with NF1.

**What happens to the results and will my taking part be kept private?**

The results of this study will be shared with health professionals, charities, support groups and people involved in research in this area. It may be reported in professional journals or at meetings and conferences.

You will not be identified or mentioned by name at any point. Your responses will be kept safe and we will not discuss you with anyone else.

**Who is supporting this study?**

This study has been approved by the Ethics Committee at the Faculty of Health and life Sciences at The University of the West of England. This means that a group of people have read through the information about the study and agree that it is being carried out in an acceptable way.

**What next?**

Thank you for taking the time to read this. If you are under 18 please ask your parent/carer to also read through this information sheet. It would be a good idea to have a think about whether or not you would like to do this. If you are interested or would like to ask any questions please call the researcher whose name is Jenny on 07500121137 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

Jenny Barke, PhD Researcher, Centre for Appearance Research at the University of the West of England.

Dr Diana Harcourt, Reader in Health Psychology and co director of the Centre for Appearance Research at the University of the West of England. Email: [Diana2.Harcourt@uwe.ac.uk](mailto:Diana2.Harcourt@uwe.ac.uk) Tel: 0117 3283967

Prof. Jane Coad, Professor in Children and Family Nursing, Coventry University.

Dr Ainsley Newson, Senior Lecturer in Biomedical Ethics at the Centre for Ethics in Medicine at Bristol University.

## Appendix 4: Information sheets for parents (interview study)

### Young Peoples' Experiences of Neurofibromatosis Type 1: An Exploration of Psychosocial Issues

#### Information Sheet for parents

You and your son/daughter are invited to take part in a research study. Before you decide whether or not you would like to take part it is important that you know why the research is being carried out and what you will be asked to do. This sheet aims to give you the information you need so that you can decide if this study is something you would like to take part in. If there are any points you are not clear on please do ask questions. Thank you for your time reading this sheet.

#### What is the purpose of the study?

Neurofibromatosis type 1 (NF1) is very variable. Some people may not even know that they have NF1. For other people some aspects such as the changes to appearance that can occur may cause more difficulty. We would like to understand how young people experience having NF1. There has not until now been much research that focuses on how NF1 impacts specifically on young people. If we can understand how young people with NF1 feel and how they manage their condition then we can try to help to make things better for other young people and their families in the future.

This study is being carried out by researchers at the centre for appearance research at the University of the West of England. This study will be part of a research degree (PhD) for one of the researchers.

We would like to ask you to think about how having NF1 may or may not affect your son/daughter. This will be used to help write questions for other parents to answer in a later study. Your son/daughter might also be deciding whether or not to take part in a separate part of the study (please see the young person's information sheet **Ref:** YPIS.S2 V2 March 11).

#### Who can take part in the study and do I have to?

You are being asked if you would like to take part as you/your child contacted the research team for more information after seeing one of our adverts. We are interested in talking to young people aged 14-24 who have a diagnosis of NF1 and their parents/carers.

You do not have to take part. It is totally up to you and if your son/daughter is under 18 we would also need your permission for them to take part. If you do decide to take part you will be asked to read and sign a consent form saying you would like to take part. If you change your mind about taking part you can stop at any time; no one will mind or be upset.

#### What will I have to do?

If you decide you would like to take part we would like you to meet with a researcher at your home or somewhere near where you feel comfortable talking at a time that's good for you. It won't be a formal interview and there are no right or wrong answers. The researcher will ask you some general questions about yourself and your son/daughter.

The interview should take about half an hour. The researcher can be contacted with queries by email, phone or text. We understand that some of this topic may be personal and sensitive and hard to talk about, it's fine if you don't want to talk about certain things and if you feel uncomfortable you can stop at any time.

We would like to tape the conversations; these will be written up and then destroyed when the study is finished. Only the researchers on this study will be able to listen to the tape and your name won't be linked to it. If you'd rather the researcher could take notes on the conversation instead.

**What are the benefits?**

We don't expect there will be any direct benefits for you; some people find it interesting to take part and to talk about their experiences. The information will help us to understand your experiences and write a questionnaire that will be used in further research. We would like to make sure we're asking the right questions about NF1 and we hope that overall this research programme will help to inform the provision of care and support for young people with NF1.

**What happens to the results and will my taking part be kept private?**

The results of this study will be shared with health professionals, charities, support groups and people involved in research in this area. It may be reported in professional journals or at meetings and conferences.

However you will not be identified or mentioned by name at any point. Your responses will be kept safe treated as confidential and we will not discuss what you tell us with anyone else.

**Who is supporting this study?**

This study has been approved by the Ethics Committee at the Faculty of Health and Life Sciences at The University of the West of England. This means that a group of people have read through the information about the study and agree that it is being carried out in an acceptable way.

**What next?**

Thank you for taking the time to read this. If you are interested in taking part or would like to ask any questions please call the researcher, whose name is Jenny Barke, on 07500121137 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

Jenny Barke, PhD Researcher, Centre for Appearance Research, University of the West of England.

Dr Diana Harcourt, Reader in Health Psychology and co director of the Centre for Appearance Research at the University of the West of England. Email: [Diana2.Harcourt@uwe.ac.uk](mailto:Diana2.Harcourt@uwe.ac.uk) Tel: 0117 3283967

Prof. Jane Coad, Professor in Children and Family Nursing, Coventry University.

Dr Ainsley Newson, Senior Lecturer in Biomedical Ethics at the Centre for Ethics in Medicine at Bristol University.

## Appendix 5: Information sheets for health professionals (interview study)

### Information Sheet for health professionals

#### Young peoples' experiences of Neurofibromatosis Type 1:

You are invited to take part in a research study. This sheet aims to give you the information you need so that you can decide if this study is something you would like to take part in. Thank you for your time reading this sheet.

**What is the purpose of the study** Despite the potential impact of NF1 on quality of life and psychosocial adjustment during adolescence there is limited research that examines the actual lived experience of young people with NF1. Therefore this research aims to explore the psychosocial impact of NF1 on adolescents' lived experience by examining both resilience and the challenges being faced, in order to identify any support needs and the factors that might help positive adjustment. The study you are taking part in is a series of interviews with specialists who work with young people with genetic conditions including NF1. This is being undertaken in order to get an insight into current provision and care for young people with NF1 and possible support needs. This study is being carried out by researchers at the centre for appearance research at the University of the West of England. This study will be part of a research degree (PhD) for one of the researchers.

**Who can take part in the study and do I have to?** Participation is voluntary, if you do decide to take part you will be asked to read and sign a consent form. If you change your mind about taking part you can stop at any time.

**What will I have to do?** If you decide you would like to take part we would like you to meet with a researcher. During the interview the researcher will ask you some general questions about your experiences working with young people/families with NF1/genetic conditions. The interview should take less than half an hour. The researcher can be contacted with queries by email or phone. We would like to record the conversations; these will be written up and then destroyed when the study is finished. Only the researchers on this study will be able to listen to the tape and your name won't be linked to it. If you'd rather the researcher could take notes on the conversation instead.

**What are the benefits?** We don't expect there will be any direct benefits for you; however the interviews will be used to write a questionnaire and results will be feedback to participants. We would like to make sure we're asking the right questions about NF1 and we hope that overall this research programme will help to inform the provision of care and support for young people.

**What happens to the results and will my taking part be kept private?** The results of this study will be shared with health professionals, charities, support groups and people involved in research in this area. It may be reported in professional journals or at meetings and conferences. However you will not be identified or mentioned by name at any point. Your responses will be kept safe treated as confidential and we will not discuss what you tell us with anyone else.

**Who is supporting this study?** This study has been approved by the Ethics Committee at the Faculty of Health and life Sciences at The University of the West of England.

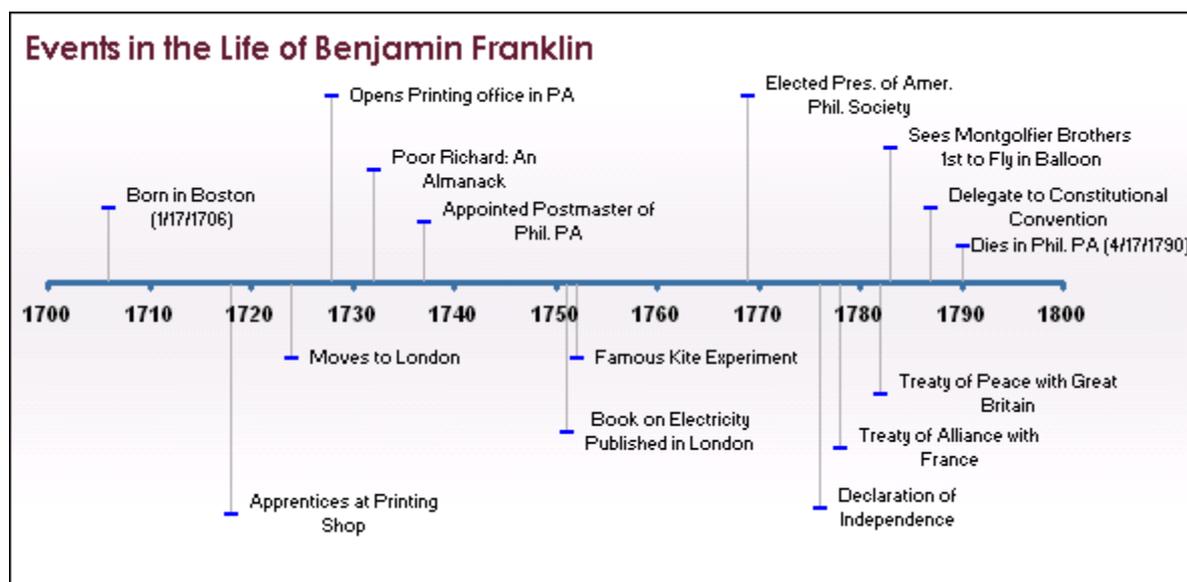
**What next?** Thank you for taking the time to read this. If you are interested in taking part or would like to ask any questions please call the researcher, whose name is Jenny Barke, on 0117 3281891 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk). Jenny Barke, PhD Researcher

Dr Diana Harcourt, Reader in Health Psychology and co director of the Centre for Appearance Research at the University of the West of England. Prof Jane Coad, Professor in Children and Family Nursing, Coventry University and Dr Ainsley Newson, Senior Lecturer in Biomedical Ethics at the Centre for Ethics in Medicine at Bristol University

## Appendix 6: Timeline Information (sent to young people prior to interviews)

### What is a timeline?

Timelines are normally used to explore events over a time period for instance the timeline below explores events in Benjamin Franklin's life.



Timelines in this project will all be about you. Before the interview you might want to think about what things in your life have been important or memorable either related to NF1 or more generally. During the interview the researcher will ask you some general questions about yourself and will ask you to make a timeline with them-this can be in words like the one above or you might want to make one using pictures.

If you want you could use copies of photos or other special items that you have, maybe a ticket from a special event, or a picture of your first day at school, feel free to bring memorable items to the interview. You don't have to do anything in advance if you don't want to, the aim is to talk about your experience of having NF1, and the timeline can be a useful way of exploring this.

The timeline will be made on a large piece of A3 paper. The timeline is yours to keep although the researcher would like to take a picture with your permission. If you want to ask any questions please contact the researcher whose name is Jenny on 07500121137 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

## Appendix 7: Interview guide young people

Before we start I would like to remind you that you do not have to answer any questions that you don't want to and if you don't want to answer a question you don't have to say why. You can stop the interview at any time if you don't want to carry on. The questions I am going to ask don't have right or wrong answers; I am interested in finding out what you feel about things. I would really like it if you could be as honest and open as you can when you are answering questions. No one else will know what you have said. Does that make sense? Do you have any questions at all?

**I would like first of all to ask you a little about yourself. (Questions could include):**

How old are you?  
 What do you do? (school, study, work)  
 First of all can I ask what do you understand by NF1?  
 Do any of your family have NF1?

**I am now going to ask you a bit about having NF1 and what I would like to do as we talk is plot on this time line different things we talk about (draw line write current age at end and have a chat about this)**

Do you remember when you first knew you had NF1?  
 Did/do you tell anyone, maybe at school or friends-at what ages?  
 How did/do family/friends react at different times?  
 Looking at the timeline what sort of medical treatment have you had at different ages?  
 Transition to adult care?

**For some people with NF1 there can be changes to the way they look, I am going to ask you a bit about how you feel about the way you look and if that has changed at all. (Questions could include):**

Sometimes how we look can seem quite important and sometimes not, how important do you think the way you look is? Have there been times that have been easier/harder?  
 Could you describe for me how you look?  
 Is there anything that you like or don't like in particular?  
 If you could change anything about the way you look is there anything you'd change?  
 Have you ever tried to change how you look?  
 Have you ever compared the way you look to other people your own age?  
 Have you ever thought that people treat you differently because of your NF?  
 In what way?

**Another area I am interested in talking about is support.**

Are there any events that you found harder because of the NF?  
 Have you had much contact with other people with NF1?  
 Can you give me an example?  
 Is there anything good about having NF?  
 What support have you had/been offered/is there available?  
 Have you ever contacted or been involved with any support groups?  
 What support would you like to have had and when?

Would you like to keep the timeline or can I take a picture of the timeline? It's completely fine if you'd rather not, it will only be seen by the research team and I would make sure your name couldn't be seen. **Is there anything else you would like to add to what you have already said?**

Once I have finished the survey I will send you a copy-not to complete but to have a look at so you can see what you have helped to design and also I would love to hear what you think of it. When I have finished and written up all my studies I will send you a summary.

## Appendix 8: Interview guide parents

Before we start I would like to remind you that you do not have to answer any questions that you don't want to and if you don't want to answer a question you don't have to say why. You can stop the interview at any time if you don't want to carry on. The questions I am going to ask don't have right or wrong answers; I am interested in finding out what you feel about things. I would really like it if you could be as honest and open as you can when you are answering questions. No one else will know what you have said. Does that make sense? Do you have any questions at all?

**I would like first of all to ask you a little about you and your family. (Questions could include):**

How many children do you have?  
 How old are they?  
 First of all can I ask what do you understand by NF1?  
 Do any of your family have NF1?

**I am now going to ask you a bit about having NF1 and what I would like to do as we talk is plot on this time line different things we talk about (draw line write current age at end and have a chat about this)**

Do you remember when you first knew your son/daughter had NF1?  
 How were they diagnosed?  
 Looking at the timeline what sort of medical treatment have you had at different ages?  
 How did family/friends react initially?  
 Has this changed over time?  
 Have there been any particularly challenging times/aspects?  
 Is there anything good about having NF?  
 Could you describe for me what it has meant for you and your family?  
 Have you ever thought that people treat your son/daughter differently because of NF?  
 In what way?

How did you tell your son/daughter about NF?

**Another area I am interested in talking about is support.**

Have you/your son or daughter had much contact with other people with NF1?  
 What support have you/your son or daughter had/been offered/is there available?  
 Have you/ your son or daughter ever contacted or been involved with any support groups?  
 What support would you/your son or daughter like to have had and when?

Would you like to keep the timeline or can I take a picture of the timeline? It's completely fine if you'd rather not, it will only be seen by the research team and I would make sure your name couldn't be seen.

**Is there anything else you would like to add to what you have already said?**

Once I have finished the survey I will send you a copy-not to complete but to have a look at so you can see what you have helped to design and also I would love to hear what you think of it. When I have finished and written up all my studies I will send you a summary.

## Appendix 9: Interview guide health professionals

- Can you tell me a little about your role and how you came to it?
- How are people referred to you, what are initial sessions like?
- After the initial session do you meet regularly or is it opt in-who books sessions?
- Are meetings with the whole family or do you see young people alone-is there an option for this or a stage when it happens?
- What are the biggest/most common concerns for (A) parent and (B) child?
- Is appearance mentioned in these concerns? (ask for examples, if the answer is no ask similar questions-do people mention how they look or that other people look at them or maybe talk about covering the fibromas?)
- When-are there particular ages? (Prompts-is bullying ever mentioned, starting school, transition to secondary school, dating, having children themselves?)
- How easy or difficult do you find it to talk about appearance issues with young people with NF1, and their families?
- How do you respond? What advice do you give families and YP?
- What support is available (at your centre and wider) for 1-yp and 2-families?
- What support would you like to see/do people ask for?
- Is there anything else that you would like to add?

## Appendix 10: Interview consent forms: young people under 18

### Informed Consent Form: young people

#### Experiences of Young People with NF1

Researcher: Jenny Barke, Centre for Appearance Research, University of the West of England

- I confirm I have read and understood the information sheet for this study
- I understand I am participating voluntarily and can withdraw at any point
- I agree to take part in the above study
- I agree to allow the interview to be tape recorded
- I understand that I can withdraw from the study at any time without any need for explanation and can withdraw my data before **5<sup>th</sup> September 2011**

.....

Name of participant	Date	Signature
---------------------	------	-----------

.....

Parent/Carer	Date	Signature
--------------	------	-----------

(If participant is under 18)

.....

Researcher	Date	Signature
------------	------	-----------

**1 for participant, 1 for parent/guardian if participant is under 18 and 1 for researcher**

**Appendix 11: Interview consent forms: young people 18 and over**

**Informed Consent Form: Young People**

**Young peoples' experiences of Neurofibromatosis Type 1:**

**An exploration of psychosocial issues**

Researcher: Jenny Barke, Centre for Appearance Research, University of the West of England

- I confirm I have read and understood the information sheet for this study
- I understand I am participating voluntarily and can withdraw at any point
- I agree to take part in the above study
- I agree to allow the interview to be tape recorded
- I understand that I can withdraw from the study at any time without any need for explanation and can withdraw my data before 5<sup>th</sup> September 2011

Name of participant	Date	Signature

Researcher	Date	Signature

***1 for participant and 1 for researcher***

## Appendix 12: Interview consent forms: Parents

### Informed Consent Form: Parent

#### Young peoples' experiences of Neurofibromatosis Type 1:

#### An exploration of psychosocial issues

Researcher: Jenny Barke, Centre for Appearance Research, University of the West of England

- I confirm I have read and understood the information sheet for this study (*ref-PCIS.S2 V2 March 11*)
- I understand I am participating voluntarily and can withdraw at any point
- I agree to take part in the above study
- I agree to allow the interview to be tape recorded
- I understand that I can withdraw from the study at any time without any need for explanation and can withdraw my data before 1<sup>st</sup> July 2011

Name of participant	Date	Signature

Researcher	Date	Signature

***1 for participant and 1 for researcher***

## Appendix 13: Interview consent forms: health professionals

### Informed Consent Form: Health Professionals

#### Young peoples' experiences of Neurofibromatosis Type 1

Researcher: Jenny Barke, Centre for Appearance Research, University of the West of England

- I confirm I have read and understood the information sheet for this study (*ref-study 1 version 2 January 2011*)
- I understand I am participating voluntarily and can withdraw at any point
- I agree to take part in the above study
- I agree to allow the interview to be tape recorded
- I understand that I can withdraw from the study at any time without any need for explanation and can withdraw my data before 1<sup>st</sup> April 2011

Name of participant	Date	Signature

Researcher	Date	Signature

**1 for participant and 1 for researcher**

## Appendix 14: NRES email regarding health professionals interviews

**From:** NRES Queries Line [queries@nres.npsa.nhs.uk]  
**Sent:** 21 October 2010 14:42  
**To:** Jenny Barke  
**Subject:** RE: Query re ethical approval

Your query was reviewed by our Queries Line Advisers.

Our leaflet "Defining Research", which explains how we differentiate research from other activities, is published at:<http://www.nres.npsa.nhs.uk/rec-community/guidance/#researchoraudit>

Based on the information you provided, our advice is that the project is not considered to be research according to this guidance. It would appear to be **service evaluation** and it wouldn't require REC review and therefore it does not require ethical review by a NHS Research Ethics Committee.

If you are undertaking the project within the NHS, you should check with the relevant NHS care organisation(s) what other review arrangements or sources of advice apply to projects of this type. Guidance may be available from the clinical governance office.

Although ethical review by a NHS REC is not necessary in this case, all types of study involving human participants should be conducted in accordance with basic ethical principles such as informed consent and respect for the confidentiality of participants. When processing identifiable data there are also legal requirements under the Data Protection Act 2000. When undertaking an audit or service/therapy evaluation, the investigator and his/her team are responsible for considering the ethics of their project with advice from within their organisation. University projects may require approval by the university ethics committee.

This response should not be interpreted as giving a form of ethical approval or any endorsement of the project, but it may be provided to a journal or other body as evidence that ethical approval is not required under NHS research governance arrangements.

However, if you, your sponsor/funder or any NHS organisation feel that the project should be managed as research and/or that ethical review by a NHS REC is essential, please write setting out your reasons and we will be pleased to consider further.

Where NHS organisations have clarified that a project is not to be managed as research, the Research Governance Framework states that it should not be presented as research within the NHS.

If you have received advice on the same or a similar matter from a different source (for example directly from a Research Ethics Committee (REC) or from an NHS R&D department), it would be helpful if you could share the initial query and response received if then seeking additional advice through the NRES Queries service.

However, if you have been asked to follow a particular course of action by a REC as part of a provisional or conditional opinion, then the REC requirements are mandatory to the opinion, unless specifically revised by that REC. Should you wish to query the REC requirements, this should either be through contacting the REC direct or, alternatively, the relevant local operational manager.

Regards

**Appendix 15: Example of coding of young person’s interview transcript**

**Interview with Sarah (age 23)**

Interview Transcript (I=Interviewer P=Participant)	Code	Subtheme	Theme
I: Good, good...and you’ve not seen, it looks like, anybody who’s a particular NF specialist			
P: Em...no, no I haven’t ever met an ‘NF’ specialist. I’ve been to Genetics...Genetic Counselling em....like a couple of years ago...	Specialist  Genetic Counselling	Healthcare	Understanding and Misunderstanding
I: Yeah, I was going to ask that, if you’d ever seen anybody. Was that here or?			
P: It was in {place name} actually so I’ve always ....it was yeah I saw a Genetic Counsellor			
I: Was that your decision to go and sort of seek that information?			
P: Yes it was, it was. Em... but it was for anything em... in particular really, it was just..... I wanted to talk to them about contraception advice. Because er.... I wasn’t sure...I went on the pill and the pill...I’ve got a Neuro-fibroma at the back of my head there and I don’t know if it was just me, but...psycho-somatic or whatever, but I felt like....I thought that it was...It had grown since I started the pill and I thought it might be because of the hormones and being er.... you know a....I was doing my {Degree} at the time; it just that I was very familiar with how to look for research and still am and there’s absol....and there’s one study that’s been done about er.... contraception in patients...women with NF and that was it and it was a very very small study but there isn’t anything there that says it’s okay really. I wasn’t reassured by it so that’s why I went to the Genetic Counsellor, just because of that (laugh)	Genetic Counselling  Information seeking  Contraception         Specialist information seeking	Healthcare	Understanding and Misunderstanding

so yeah there's loads of things going on.			
I: Oh where they helpful?			
P: Em...not...they didn't tell me anything I didn't already know, they just checked my spine and that kind of thing, you know. While I was there, they thought they may as well check that kind of stuff, but they didn't know. I mean when I talk about these kinds of things with Healthcare professionals, because they see me as the 'expert' I think...I come away from there without any real advice...and they just kind of tell me that I should...it's not bad of them, it's good, that they kind of give me this empowerment and whatnot, but I come away from there just with the same kind of thoughts and stuff that I went in there with, I never really had guidance or anything.	Being the expert  Finding information	Healthcare	Understanding and misunderstanding
I: Yeah, do you think that's your education?			
P: It was my {Identifiable details of qualifications} when I...I don't ever like go into a doctor's office and tell them this, I kind of...but they...I do talk about the fact that I know what's there. 'Cos then I don't want them to...I want them to be able to speak to me on my level kind of thing and understand everything that I have in my head em... and why I'm worried and the reasons why I'm worried is because the scientific literature is so lacking and I mean that's why you're doing this PhD and em....	Being the expert  Not a well known condition	Healthcare	Understanding and misunderstanding
I: Yeah...			
P: And er...but then that means they don't have any answers either really.	Not a well known condition	Healthcare	Understanding and Misunderstanding
I: So in terms of.... you've sort of always known that you've had NF have there been points....can you remember anyone ever talking to you and saying this is what it is, or has it been more of a...a			

slow....?			
<p>P: It's er...well when I was diagnosed when I was four...em...my Mum doesn't really; she's not...er...very well educated and stuff and she er...didn't know much about it herself and like for the first, I don't know how many years of my life I was saying 'Neuro-fibromatosis' wrong. I don't know what I was calling it. Something else, because my Mum had been calling it something else, so obviously, didn't really know much about it and stuff. But I remember when I was er...quite young...they must have given ...the doctors must have given my Mum leaflets when I was about four or five but we kept them in the house and from a really young age probably like, I don't know, whenever I was able to read I remember reading these leaflets and I had an interest in it so I kind of knew a little bit about what it was and not really. And it was when I was about fourteen I think, em...when I really starting kind of going online and reading about it, so everything I knew about it I found out for myself and that's when I understood em.... what it actually was.. em...because for me it had always been a leg thing. Em...and then to kind of reconceptualise that it was quite weird to think that there's somebody else with NF who is affected in a completely different way and em.... so yeah up until that age sort of thirteen fourteen, it wasn't...it was just...I've got NF, that means my legs and my shin bones don't work properly. So...there's been a lot of kind of learning since then.</p>	<p>Diagnosis and treatment</p> <p>Not knowing what NF1 is</p> <p>Parental knowledge</p> <p>Interest growing during adolescence</p> <p>Information seeking (Internet)</p> <p>Reconceptualising</p>	<p>What is NF1?</p>	<p>Different things to different people</p>
<p>I: When you started looking, you said you were looking on the internet, were there any bits that were useful and horrible or....?</p>			
<p>P: Em....</p>			
<p>I: Was there anything in particular you remember looking at?</p>			
<p>P: Well I went...I used to go on</p>	<p>Finding information-</p>	<p>What is</p>	<p>Different things to</p>

<p>like forums and stuff and that was really helpful, em...and I talked to people. But I remember finding out, well there's first of all, the thing about the Elephant man which is just kind of a really common thing that people raise. And it was on...I can't think where I first heard about 'oh you know, Elephant Man had NF' but I remember finding out quite soon after I'd heard that, that he actually...that John Merrick, is that his name? Didn't actually have NF, it wasn't confirmed em...but then the thing that was really horrible that I found out. At the time it was anyway. Em...was about the impact of NF on my ability to have children. So I found out about that when I was about thirteen, fourteen and that really upset me at the time so I was instantly trying to find a solution and then started reading about pre-implantations genetic diagnosis and stuff so.... just to kind of em....at that age, all of a sudden think will you...that whole stereotype of growing up and getting married and having children isn't going to happen for me in that way. Em...that was horrible. But because I kind of found an answer for myself and I had friends, really close friends that I talked to about it, it wasn't...I feel like it wasn't a massively tragic moment or anything, it was just something that was a little bit of, something that I had to carry with me kind of thing.</p>	<p>Internet</p> <p>Elephant Man</p> <p>Having children</p> <p>Genetic information</p> <p>Reconceptualising</p>	<p>NF1?</p> <p>What does NF1 mean for me?</p>	<p>different people</p>
<p>I: Yeah. Is that something that you've talked about with sort of Genetic Counsellors, or anyone that you've seen. Or has it been more...</p>			
<p>P: No when I went to the Genetic Counsellor, I talked to them about that and well, it was good because he actually.....er..... well he kind of, er... he made it seem like I was... 'cos I always thought I...worst case scenario is that you're own er...NF has no bearing on the....kind of NF your child will have so even though I feel like I've got it relative....the way its impacting me is relatively mild, em.... I was kind of...I thought about the worst case scenario</p>	<p>Genetic counselling</p> <p>Expert advice</p>	<p>Healthcare</p>	<p>Understanding and misunderstanding</p>

<p>which was that you know, any child I have could be born without limbs; could be born with severe learning disabilities, but the Genetic Counsellor gave me a statistic or something which was like a...the risk of em.... of the child having it really...having it you know really badly. Em... I can't remember what that was but I took some comfort from that because it made me think well it's not gonna be definitely bad.</p>	<p>Information into perspective</p>		
<p>I: Yeah.</p>			
<p>P: But I mean at the moment I'm....don't want children anyway so if....so now for me, this isn't a problem. Whereas, when I was a bit younger it was more of an issue.</p>	<p>Having children (decision making)</p>	<p>What does NF1 mean for me?</p>	<p>Different things to different people</p>
<p>I: Yes I can understand that. You were saying that you talk to friends. Do you think your NF was visible to people and in what way sort of when you were younger</p>			
<p>P: Em...it's always been visible because of my legs and because when I was young...er. yeah when I was four that was another one. I had to have an operation in order to set....in order to kind of get my shin bone set into place. So I've always had scars and then because....I can't remember why they did this, but because of the problems with my right leg, because that where all the problems have been, I had an operation on my left leg as well so I've always kind of walked with a limp and had scars and that's the way it's been visible and its....so I mean doing PE and stuff at school, at Primary school and what have you.....and High School and stuff, it was always visible because of shorts and stuff and I had to wear splints as well when I was much younger, so little support things and also used a wheelchair for a while at Primary school because both my legs were a bit dodgy. Em... so it was always visible and yeah.... even if it was...even if my kind of scars and stuff were covered up it was visible to people that I was different</p>	<p>Medical treatment (description)</p> <p>Medical treatment Description</p> <p>Impact/symptom</p> <p>Education (PE)</p>	<p>What is NF1</p>	<p>Different things to different people</p>

because I had like splints and was walking on crutches or in a wheelchair or whatever.	Impact/symptom		
I: And how do you think friends and family have dealt with that. Has that been something that's been....that you felt quite comfortable about talking with people? Or...do you feel that people sort of judge you based on the way you look. At school....?			
P: Do you mean early on?			
I: Yeah sort of earlier on, did you feel a sense of sort of being judged for the way you looked, sort of back in Primary school, or...?			
P: Not really. In Primary school I remember, I still think about it now and I'm just like really surprised that I wasn't ever bullied because of it. Em...but I wasn't ever made to feel....judged and stuff. I think because we didn't....I never really knew what NF was and my Mum didn't know, so the teachers didn't know so it was just 'something's wrong with my legs', so that was always kind of known throughout my classmates and my teachers and er...it was quite open. 'Cos I remember I had to leave school a couple of times for ages and go and have operations and stay in Surrey for weeks at a time and then be off school for weeks and they always did a little thing in like assembly where they would give me a going away present or whatever and it was always a little bit like embarrassing, but obviously I liked it because I was getting loads of free stuff..	Changes over time  Reconceptualising  Attention (good and bad)	What is NF1?	Different thing to different people
I: Presents...yeah (laugh)			
P: Yeah. Attention. Em...so I never felt like I couldn't talk about it with people and I never felt like people didn't understand. Em...it was just something that was there. It was never really like a massive deal. Em, I don't remember it being anyway. Em... 'cos I was....I was like,...I went to a really	Primary friendships positive	Friends	Relationships and reactions

<p>rubbish Primary school like er... in {name} and I was not.... I'm not like blowing my own horn or anything, but I was kind of one of the top students so that was thing. (laugh) That was kind of my identity was that I was like going to do well and I went to a Grammar school and stuff....the leg thing was just kind of....secondary to that so I wasn't ever judged.</p>	<p>Identity</p>		
<p>I: Yes. What about Secondary School? Was that the same or.....?</p>			
<p>P: Secondary school was... [pause] I don't know. I remember it took me a long time to feel like I fit in and I think I feel a little bit kind of like that all the time around people and I think it is a lot to do with the fact that I'm different in that I...I look different because of the legs and I walk differently because of the er...operations I've had and stuff. Em...and I ended up having a really good group of friends which is....within the first year or so, so that was fine and I never really felt judged by anybody. Em [pause]...</p>	<p>Social changes Socially anxious  Good friends</p>	<p>Friends</p>	<p>Relationships  And reactions</p>
<p>I; Your group of friends that you had, did they know from day one....well not from day one, but did they know you have NF, this is..... and I wonder did they think that was a 'leg thing'? You know did they...?</p>			
<p>P: Yeah, I think I probably would have explained it to them. Em...I can't remember.... I've got a really vague memory that at some point I talked to the class about it or something, but em...they would have thought .... at first they would have thought it was a leg thing because that's how I thought of it up until like I say, thirteen or fourteen, so up until I was in year nine or something...em...so they would have thought that. But then...but when I went....when I went to the School, High School I was kind of alright. My....I'd gotten over a lot of the operations and stuff; didn't have to use any...didn't have to use a wheelchair, didn't have to use er</p>	<p>Explaining NF1  Impact (social and health)</p>	<p>Friends</p>	<p>Relationships and reactions</p>

<p>crutches. I wasn't walking with as much of a limp as I do now....;'cos that was before like. I had a big operation when I was like er....seventeen...</p>			
<p>I: Yeah er...</p>			
<p>P: Right back there....and...em....so I don't think it was that visible to people really and I think people mentioned the limp and I used to get annoyed at that because I....I never really thought about my own limp and when the....and when people....I remember one girl talking about it, mentioning it to me and I just got annoyed at the fact that she'd even raised it because er.....[pause] I guess....it was a bit rude of her but also it was a bit like....of a...horrible thing to confront for me because.... 'cos in my mind at that point I was kind of alright and normal and stuff so....er....then when it came to thing like scars and stuff em...I never felt judged or anything for that. Not really. Em...</p>	<p>Visibility</p> <p>Managing others questions</p> <p>Impact of questions</p>	<p>Other people</p>	<p>Relationships and reactions</p>
<p>I: And what about, you were saying that thirteen; fourteen you started to understand a bit more and you said about having early puberty as well...did you have sort of many fibromas appearing at that point? Was that.....?</p>			
<p>P: Em...I don't remember them appearing then, but over the years from about then onwards I've noticed them and like....I don't have.... I've...it's always really horrible when you see like oh you know, man with 20lb tumour and whatever and...in the Metro and on Bodyshock 'cos you know instantly that persons got NF and its horrible and then....so...but I know that it's not what I had....isn't bad at all, but I've started realising that little bits about my appearance that I don't like are because of NF, so I've got like...there's this one here; I've got like one on my arm somewhere. My arm there and stuff.... and they're all dotted around like that em.... so it was from that age that I started realising all of this stuff is because of the NF. Like the reason why</p>	<p>Representations of NF on TV/Internet etc</p> <p>Appearance concern/awareness</p> <p>Reconceptualising</p>	<p>Media</p> <p>What does NF1 mean for me?</p>	<p>Understanding and misunderstanding</p> <p>Different things to different people</p> <p>Different things to different people</p>

<p>I'm short em...I've got a slightly larger head than average. Hats don't fit me and stuff. But em...yeah...that's when I kind of started noticing all these little things and things like the birthmarks and stuff. Because for me it was always really normal, and I guess 'cos my Dad had lots as well, I never really thought, having lots of birthmarks is a bit, is a bit weird, but...'cafe au lait' spots, but em...then I started thinking oh, you know.....this is just something that I have and people always ask about 'oh what happened to your arm?', and 'what's that on your head?', 'cos I've got birthmarks and they always think it's like bruises and stuff and that's when I started thinking, oh you know, this is annoying you know, 'cos it's something....it's all because of NF. And its things that other people don't have and that I don't like about myself and that are being pointed out to me.</p>	<p>Reconceptualising</p> <p>NF1 normal in family</p> <p>Feeling different</p> <p>Answering questions</p>	<p>Family</p> <p>Friends/other people</p>	<p>Relationships and reactions</p> <p>Relationships and reactions</p>
<p>I: When somebody says that to you, and says 'what's that?', do you have an answer. Do you...how do you answer that question? Do you, are you quite happy saying, well its NF, or do you...?</p>			
<p>P: Not....well I just kind of....if people....people don't ever really ask er.... the one that I've got a couple of times is 'oh what's happened to your arm?', 'cos...I'll show you, I mean it's not even that bad but I've just got like a couple of birthmarks and stuff there and from far away people always think they're bruises for some reason and I just say oh they're just birthmarks. I don't ever really explain because I don't feel like it's necessary really. If someone was to ask me a more.....er.... in-depth...if someone was to ask me about my legs, then I would probably go into it a little bit more, but the superficial kind of stuff I just...I don't really talk about in detail. I just want to change the subject.</p>	<p>Managing others questions</p>	<p>Other people/friends</p>	<p>Relationships/reactions</p>

**Appendix 16: Example of coding of parent’s interview transcript  
(Interview with Gwen)**

Interview Transcript: ( I=Interviewer P=Participant)	Code	Subtheme	Theme
I: There that one’s on. Um ok sorry, so you have 3 children?			
P: Yes...	Background		
I: ...and it’s your youngest...			
P: ...V is the youngest...	Background		
I: ...and it’s V who has NF there’s no-one else in the family?			
P: No, no other member and either side down the history line we couldn’t find anyone so they told us it was a new mutation what is it?	Background		
I: Yeah mutation or sometimes they call it De novo sometimes I’ve heard both used.			
P: Yes.			
I: So when was diagnosis then?			
P: Um from the age of 6 months they discovered that V wasn’t normal, what they called normal, tests were carried and she was quite floppy but it wasn’t until she was about a year and a half to three years before they come up with... at the age of a year and a half they suspected NF which I had never heard of and then when she was about 3 it was diagnosed that that's what she had because of the freckles they felt...	Initial diagnosis  Time taken to diagnose	diagnosis	Information and support
I: How was that for you?			

<p>P: Um I didn't really worry at that stage because I could not see any lumps or tumours it was just these birth marks and I thought which was fine but then when I went on the internet and read up at the .....which is a wrong thing to do because you don't know who wrote these and I was reading too much into it and then I would get quite upset is this what her life's going to be. So there is no two people the same they are all individual and they'll all be different and as V was getting older you realise that you have to learn with it, face it as it comes.</p>	<p>Symptoms and concerns</p> <p>Information seeking-internet</p> <p>Too much information</p>	<p>Diagnosis</p> <p>Challenges faced by the parent</p>	<p>Information and support</p> <p>Challenge</p>
<p>I: Absolutely so the diagnosis who was in charge of the diagnosis were you going through GP's and different doctors or...?</p>			
<p>P: Um V was only a week old when she was first admitted to hospital so that was our first um... she was put into hospital when she was a week old. Following that it was the midwife came out and she was referred not back to the GP but referred to Paediatrician in the hospital so basically that's where it started with V {hospital} with the Paediatrician not her GP. She hardly knew her GP it's mostly the hospitals and the consultants and then she's referred to {Name} as you have already heard...</p>	<p>Early medical concerns</p> <p>Professionals seen</p>	<p>Diagnosis</p> <p>Healthcare</p>	<p>Information and support</p>
<p>I: ...and that's in the specialist clinic?</p>			

P: Yes, yeah...			
I: ...and when did that start going to specialists?			
P: Um as I said they told us when she was three that she had NF then a wee pea dot lump appeared on her cheek, then this developed and by the time she was five the eye was closed it was quite big so I was attending the {hospital} and I said {identifiable} please do something for my child, her eye was practically closed. So they decided then they would send her {name} because there was nothing in {name} relating to the tumour at least this type of tumour...	Diagnosis and symptoms  Getting the right treatment and accessing experts	Healthcare  Challenges faced by the parent	Information and support  Challenge
I: ...nothing for...			
P: ...and I thought a tumour I automatically thought cancer things like that but then it was a benign tumour so I got a wee bit calm about that but then {name/identifiable department}	Understanding NF1  Managing treatment and family	Healthcare  Challenges faced by parent	Information and support  Challenge
{Discussion of challenges of a particular department} identifiable			
I: That's good, that's quite a lot to manage?			
P: Well it was a shock and again went to the library and researched it on the internet and it scared me because I didn't know what to expect and I thought oh dear is all this going to happen but I am more relaxed about it, it may not happen but at least I know what to do for the signs if it can happen you know what to look for. So I am at that	Information seeking  Managing information  Changing understanding and management	Diagnosis	Information and support

stage where I know quite a bit.			
I: Yeah and have you at any point during that seeing anybody like a genetic counsellor?			
P: Yes the first we attended the City Hospital for genetic counselling just to test us to see where this came from, so we both got tested and we got our blood frozen and it was discovered that it wasn't from either and even going down the histories this was just a new thing that happened in the family. {identifiable}	Genetic counselling  Genetic testing	Healthcare	Information and support
Discussion about clinics			
I: ...and when you saw the genetic counsellor were they any help in pointing you in directions and giving you advice?			
P: No well not with NF they were just able to tell us that she did not get this from either side...	Genetic counselling	Healthcare	Information and support
I: ...so it was more sort of informative and genetic diagnosis kind of thing?			
P: Yes it was just basically the time to find out where it came from Um in terms of NF there was nothing here for it but now it's changed they've set up a clinic, an NF clinic 17 years ago that wasn't there...	Lack of specialist care	Healthcare	Information and support
Discussion about clinics			
I: Have you had many situations like that where you felt the NF has impacted on V....			
P: Um in um again V had this facial disfigurement well because of that we needed to attend consultants	Appearance Getting the right	Healthcare	Information and support

<p>here and the consultants here said we will look at the signs before we send you over to {place}, we won't do it but we will send you over to {place}, so this particular, he's a cancer specialist and he looked at V and he said oh um I am cancelling that operation, she was due to go for surgery in {place} to get the tumour removed from her face, I'm cancelling that operation, and why's that, he said because there's a large tumour in V's neck and it's the size of a sausage and I says well I'm attending {place} and I think they are making a good job but he was giving me wrong information that I shouldn't be going to {place}, he should be dealing with it but he was only a cancer specialist he wasn't an NF specialist and he cancelled that operation and {place} said look come over anyway and we'll have a look and we will do our own MRI scan. I mean he did his own MRI scan and he couldn't find any tumour and the {place} was wrong again it doesn't work if you are getting outside people who don't specialise in NF they took V they need to know, there's a difference between cancer and NF.</p>	<p>treatment</p> <p>Specialists and experts</p> <p>Difficulties with specialists</p> <p>Different information</p> <p>NF experts needed/trusted</p> <p>Not trusting non NF professionals</p>	<p>Challenges faced by parent</p> <p>Healthcare</p>	<p>Challenge</p> <p>Information and support</p>
<p>I: Hmmm....</p>			
<p>P: So {place} was right so it knocks me back so nobody over here is going to touch her now because they got it wrong so I am happy with {place} and I'll continue attending {place}. So he was a top man in the cancer unit, but he was wrong.</p>	<p>Not trusting non NF professionals</p>	<p>Healthcare</p>	<p>Information and support</p>
<p>Further discussion of different clinics</p>			

<p>I: ...and as a parent have you had to deal with many comments from other people?</p>			
<p>P: Yes it has from she was a child it didn't bother V because V was pretty young, it bothered me and I wanted to say it's not her fault she is like that and sometimes I did, sometimes I kept quiet and I got stared at and I looked over at them and not said anything but my stare was saying what are you looking at her for and then I would have come out. V see that photograph where she is holding the wee bird and that was actually on holiday in Majorca and this woman was walking past and she done a double turn and she actually came straight to me...</p>	<p>Appearance not a concern when young for child</p> <p>Others staring at child</p>	<p>Childs feelings about their appearance</p> <p>Others reactions to child's appearance</p>	<p>Appearance</p>
<p>I: ...it's gorgeous...</p>			
<p>P: ...yeah that was in Majorca, she actually came to me and said um you don't mind me asking what's wrong with her eye and I said I do mind you asking because I thought you are not concerned you're nosey you didn't want to get to know me first or get to know L she just walked past and walked back and I thought she was rude and I told her yes I did mind her asking, there's a way to break it you just don't come to somebody in the street and ask what's wrong with you, that's my feeling I wouldn't do that with anyone, I would get to know a person first and if they want to share it fair enough if they don't let them be...</p>	<p>Others questions</p> <p>Intrusive questions</p>	<p>Others reactions to child's appearance</p>	<p>Appearance</p>
<p>I: ...yeah and have you had a lot of people like that...</p>			

<p>P: ...and I thought cheek she was a wee busy body and the way her whole attitude came across I thought no I am not going to share my daughter's illness with you the other person people would say to me well that's a wrong way you can only learn people but I say my learning was to let her wrong that was wrong the way she approached me and I said you don't approach I am on holiday and I don't want that. So with that I did get angry and another time was when V came out of, believe it or not it was a porter when V came back from theatre she was happily bandaged up and the porter was wheeling and he said who done that to that child and he was a porter in the hospital and the nurse joking said her dad, her dad beat her up she was a bad girl she got beat up and do you know he actually believed that and wouldn't look at him do you remember that time?</p>	<p>Intrusive questions</p>	<p>Others reactions to child's appearance</p>	<p>Appearance</p>
<p>I: What do you do now ?do you feel you cope with circumstances more...</p>			
<p>P: Um it would depend on the circumstances I would say to V, V that child's quite young and doesn't understand but I always drum it in to my own children look once and turn away don't look back that's when you cause problems, you look once and you turn away and you look back that's wrong because if someone walked in your room even myself I am going to look but I'd turn away and go to help that person but I won't look back and that is what I feel it's when the problem starts and I told V that they don't realise that</p>	<p>Explaining people's reactions to child</p> <p>Responding to visible difference</p> <p>Explaining people's</p>	<p>Others reactions to child's appearance</p>	<p>Appearance</p>

<p>they are looking twice and they shouldn't do that and then I would say maybe to a child please don't stare at V and have done that because she was getting upset about it but no she is able to handle herself and I would gently, it depends on the age and things like that but um early days I wasn't good I was quite cheeky, I'd call myself cheeky I would I would be very defensive what are you doing don't stare at my child she doesn't deserve that.</p>	<p>reactions to child  Intervening in others reactions</p>		
<p>I: You got angry.</p>			
<p>P: Yes I was very angry and defensive and I didn't like it.</p>	<p>Feeling angry</p>	<p>Challenges faced by the parent</p>	<p>Challenge</p>
<p>Participant fetches photo album and shows pictures, explains V has to do regular physiotherapy</p>			
<p>P: ...I thought V you are so beautiful no-one is going to stare at you anymore but then it grows back so you have that in between period where theres no starting and then it comes back and V says they are noticing a difference and I try to tell V to get a pen and bit down on the pen to strengthen her lip muscles to try and get the mouth closed and I would say to V they are not staring at your cheek your cheek's fine it's the mouth open and if she tries to concentrate I am trying to help her bit down on the pen or the straw what they have been asking you to do you will be exercised and her and I have wee talks and I tell V do look in the mirror but she doesn't like mirrors and if she gets new clothes she loves new clothes but won't go and look at them on her and that's what she wants to do then that's</p>	<p>Changes to appearance  Physio, treatment  Mirror New clothes</p>	<p>Child's feelings about their appearance</p>	<p>Appearance</p>

<p>fine but I tell her that um look in the mirror and we all tell her, tell her she is pretty but she's two older sisters and V wants to be like them and she does have her moments where she will, she calls it the lump, she says I wish I didn't have this lump and she would ask me why have I got this lump and things like that. Then we explain the procedures that will happen and why it's happening.</p>	<p>Mirror Lumps  Manging appearance changes</p>		
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**Appendix 17: From themes to survey questions, Young People**

Theme	Key aspects	Main points to take forward	Questions/measures on questionnaire (including page no)
Different things to different people	Perceptions of NF1	<p>What is the impact of NF1 and how do young people conceptualize the condition?</p> <p>Where do people find information and is this easily accessible?</p>	<p>Brief IPQ (Page 6-7) and 1 open ended question: "For me NF1 is....."</p> <p>Question with tick boxes-"Where do /did you find information about NF1?" plus open ended question asking for comments on information</p>
	Appearance	<p>How do people feel about their appearance?</p> <p>How noticeable do people feel their condition is?</p> <p>Is appearance young people's greatest concern?</p>	<p>Body Esteem (appearance sub scale)</p> <p>"Do you think your NF1 is noticeable to other people?" (yes/no) (plus text box for comments)</p> <p>Open ended question; "My main concern about NF1 is....." (page 7) and "For me NF1 is..."</p>
Relationships and Reactions	Social situations and others responses	<p>Does inheritance status impact on social experience?</p> <p>Are family and friends an important source of support?</p>	<p>Does anyone else in your family have NF1</p> <p>Covered in questions re support</p>
		<p>Are social situations and interactions with others important to experience?</p>	<p>Perceived Stigma Questionnaire</p> <p>Social Comfort Questionnaire</p>
Understanding and misunderstanding	Perceptions of NF1	<p>Do young people feel medical professionals know about and understand NF1?</p> <p>What aspects of healthcare experience are highlighted as important?</p>	<p>Quotes re health professionals knowledge and understanding of NF1 respondents asked to agree/disagree</p> <p>Open ended question - is anything else to add re health professionals</p>
		<p>What was educational experience?</p>	<p>Quotes re school – respondents asked to agree/disagree</p>
		<p>Where do young people look for support, what sources are most useful and what would they like to see available?</p>	<p>Where do you look for support? series of sources to tick, plus open ended question</p>
		<p>How do young people feel about awareness of NF1 and its portrayal in the media?</p>	<p>Quotes re media – respondents asked to agree/disagree</p>
Across themes	General Wellbeing	<p>Are young people generally happy?</p>	<p>Subjective Happiness Scale</p>

## Appendix 18: Information sheets and survey for young people

**Measures used (B-IPQ, SHS, BE Appearance PSQ and SCQ) are indicated in capital letters and underlined for the purpose of this appendix-this did not appear on surveys used in the study.**

You are invited to take part in a research study. Before you decide whether or not you would like to take part it is important that you know why the research is being carried out and what you will be asked to do. This sheet will give you the information you need so you can decide if this study is something you would like to take part in. If there are any points you are not clear on please ask. Please read the sheet carefully; if you are under 16 please share this information with your parents/carers as we will require their permission for you to take part. Thank you for reading this sheet.

### What is the purpose of the study?

We would like to understand how young people experience having NF1. There has not until now been much research that focuses on how NF1 impacts specifically on young people. If we can understand how young people with NF1 feel and how they manage their condition then we can try to help to make things better for other young people and their families in the future.

This study is being carried out by researchers at the Centre for Appearance Research at the University of the West of England, in Bristol. This study will be part of a research degree (PhD) for one of the researchers, Jenny Barke.

### Who can take part in the study and do I have to?

You do not have to take part. It is totally up to you and, if you are under 16, your parent/carers. If you do decide to take part completing the survey will be taken as consent to participate if you are under 16 we would also need your parent/carers consent. If you change your mind about taking part you can stop at any time; no one will mind or be upset.

Your parent/carer may also decide to take part in this study by completing the separate survey for parents. You and your parent do not have to both take part, surveys can be completed by you, your parent or you can both take part.

### What will I have to do?

If you decide you would like to take part we would like you to complete the young person's questionnaire enclosed, it is about your experience so there are no right or wrong answers.

We understand that some of this topic may be personal and sensitive; it's fine if you don't want to answer any questions and if you feel uncomfortable you can stop at any time.

There is also an online version of the questionnaire which can be found at [link](#) please feel free to complete either version.

### What are the benefits?

We don't expect there will be any direct benefits for you; some people find it interesting to take part in research and to think about their experiences. The information will help us to understand your experiences and we hope that overall this research will help to inform the provision of care and support for other young people with NF1.

**What happens to the results and will my taking part be kept private?**

The results of this study will be shared with health professionals, charities, support groups and people involved in research in this area. It may be reported in professional journals or at meetings and conferences.

You will not be identified or mentioned by name at any point. Your responses will be kept safe and we will not discuss you with anyone else.

**Who is supporting this study?**

All research in the NHS is looked at by an independent group of people, called a Research Ethics Committee. Research Ethics Committees (RECs) safeguard the rights, safety, dignity and well-being of people participating in research in the National Health Service. They review applications for research and give an opinion about the proposed participant involvement and whether the research is ethical. This study has been reviewed and given favourable opinion by NRES Committee South West – Frenchay.

**What next?**

Thank you for taking the time to read this. If you are under 16 please ask your parent/carer to also read through this information sheet. It would be a good idea to have a think about whether or not you would like to take part in this research. If you are interested or would like to ask any questions please call the researcher whose name is Jenny on 07500121137 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

As a thank you for your time completing the survey you can choose to be entered into a prize draw with a chance to win £25 worth of online or High Street shopping vouchers. If you would like to be entered please return your email address in the envelope marked 'young person'.

Jenny Barke, PhD Researcher, Centre for Appearance Research at the University of the West of England.

Supervisor: Dr Diana Harcourt, Reader in Health Psychology and co director of the Centre for Appearance Research at the University of the West of England. Email: [Diana2.Harcourt@uwe.ac.uk](mailto:Diana2.Harcourt@uwe.ac.uk)  
Tel: 0117 3283967

**Young peoples' Experiences of NF1: Survey (available online and as a questionnaire booklet)**

You are invited to take part in a research study. Before you decide whether or not you would like to take part it would be a good idea to have a think about whether or not you would like to take part in this research. If you would like to ask any questions please call the researcher whose name is Jenny on 07500121137 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

**Please note completing this questionnaire will be taken as your consent to participate**

If you are under 16 please talk to your parent/carer and make sure they are happy for you to take part, if they are happy could they please sign below:

I confirm I have read the information sheet provided and I consent to my son/daughter taking part in this study

Parent/Carers signature.....

Date.....

Please return the completed questionnaire in the envelope provided. You do not need a stamp.

**CREATE YOUR OWN ID CODE**

You have the right to withdraw from the study up to four weeks after you have completed the survey. Should you wish to withdraw, you will need to inform us by email [Jenny.barke@uwe.ac.uk](mailto:Jenny.barke@uwe.ac.uk) quoting your unique ID code. This will enable us to identify all the material that needs to be deleted due to your withdrawal from the project.

**To create your ID code please enter the first three letters of your first name and the date (day of the month) you were born.**

*If your first name is Karen and you were born on the 5th June you would enter KAR05*

Please enter your code in the box below:

**A: About you**

Are you?

- Male
- Female

How old are you?

- 24
- 23
- 22
- 21
- 20
- 19
- 18
- 17
- 16
- 15
- 14

How would you describe your ethnic group?

White

- English / Welsh / Scottish / Northern Irish / British
- Irish
- Gypsy or Irish Traveller
- Any other White background please write in \_\_\_\_\_

Mixed / multiple ethnic groups

- White and Black Caribbean
- White and Black African
- White and Asian
- Any other Mixed / multiple ethnic background please write in \_\_\_\_\_

Asian / Asian British

- Indian
- Pakistani
- Bangladeshi
- Chinese
- Any other Asian background, please write in \_\_\_\_\_

Black / African / Caribbean / Black British

- African
- Caribbean
- Any other Black / African / Caribbean background, please write in \_\_\_\_\_

Other ethnic group

- Arab
- Any other ethnic group, please write in \_\_\_\_\_

Or

- prefer not to answer

Are you in education or employed?

- School
- College
- University
- Working Full time
- Working part time
- unemployed
- other (please explain) \_\_\_\_\_

Which area do you live in?

- England
- Scotland
- Wales
- Northern Ireland
- Ireland
- Outside UK and Ireland please say which country below \_\_\_\_\_

**B: Diagnosis and Treatment**

Do you have Neurofibromatosis Type 1 (NF1)?

- Yes
- No

Roughly how old were you when you were first diagnosed?

Does anyone else in your family have NF1?

- Yes -if yes please explain what relation they are to you \_\_\_\_\_
- don't know
- No

What health professionals do you see for your NF1? (*please tick*)

- My GP
- Genetics specialist
- NF1 Specialist
- Children's doctor or nurse
- Dermatologist (skin specialist)
- Eye Specialist
- Don't know
- Other (please explain) \_\_\_\_\_

Has this changed-did you maybe see different Doctors about your NF1 when you were younger?

- Yes
- don't know
- No

How much do you agree or disagree with the following statements? (*please tick*)

Doctors and Nurses generally know about and understand NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I sometimes have to explain to Doctors and Nurses what NF1 is	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Most Doctors and Nurses don't know about NF1, I have to be the expert	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Is there anything else you would like to tell me about your experience with Doctors and Nurses and NF1?

**C: Your experience and understanding of NF1**

Please complete the following sentence in whatever way you think best describes your experience of NF1

For me NF1 is.....

For the following questions please circle the number that matches what you think **(BRIEF IPQ)**

How much does NF1 affect your life?

0      1      2      3      4      5      6      7      8      9      10

No affect at all

severely affects my life

How long do you think your NF1 will continue?

0      1      2      3      4      5      6      7      8      9      10

A very short time

Forever

How much control do you feel you have over NF1?

0      1      2      3      4      5      6      7      8      9      10

Absolutely

extreme amount

no control

of control

How much do you think treatment can help NF1?

0      1      2      3      4      5      6      7      8      9      10

Not at all

Extremely helpful

How much do you experience symptoms from NF1?

0	1	2	3	4	5	6	7	8	9	10
No symptoms at all										many severe symptoms

How concerned are you about NF1?

0	1	2	3	4	5	6	7	8	9	10
Not at all concerned										extremely concerned

How well do you feel you understand NF1?

0	1	2	3	4	5	6	7	8	9	10
Don't understand At all										Understand very clearly

How much does NF1 affect you emotionally (eg does it make you angry, scared, upset or depressed?)

0	1	2	3	4	5	6	7	8	9	10
Not at all affected emotionally										extremely affected emotionally

Please list (most important first) the 3 most important things that you believe caused your NF1.  
The most important causes for me:-

1

2

3

Please complete the following sentence

<p>My main concern about NF1 is....</p>
---

**D: What NF1 means for you (SUBJECTIVE HAPPINESS SCALE)**

For each of the following statements and/or questions, please show where on the scale best describes you

in general I consider myself:

1	2	3	4	5	6	7
Not a very			a very			
happy person			happy person			

Compared to most of my friends, I am

1	2	3	4	5	6	7
Less happy			more happy			

Some people are generally very happy, they enjoy life regardless of what is going on, getting the most out of everything. How much is this like you?

1	2	3	4	5	6	7
Not at all very			a great deal			

Some people are generally not very happy. Although they are not depressed they never seem as happy as they might be. How much is this like you?

1	2	3	4	5	6	7
Not at all very			a great deal			

Do you think your NF1 is noticeable to other people?

- No
- Yes

If yes please briefly describe how:

How much do you agree with the following statements? (please tick) (**BODY ESTEEM APPEARANCE SUBSCALE**)

I like what I see when I look in the mirror	<input type="radio"/>				
I'm looking as nice as I'd like to	<input type="radio"/>				
I'm pretty happy about the way I look	<input type="radio"/>				
I like what I look like in pictures	<input type="radio"/>				
I wish I looked like someone else	<input type="radio"/>				
There are lots of things I'd change about my looks if I could	<input type="radio"/>				
I wish I looked better	<input type="radio"/>				
My looks upset me	<input type="radio"/>				
I feel ashamed of how I look	<input type="radio"/>				
I worry about the way I look	<input type="radio"/>				

**E: How do other people treat you?**

During your normal day, you probably see and talk to many different people. We want to know how often people act in certain ways towards you. For each question, rate how often people do certain things. Make your ratings about how people treated you over the last year. First, for each question, say how often people do certain things using the scale below, by ticking **never** to **always**

**(PERCEIVED STIGMATIZATION QUESTIONNAIRE)**

1. People are friendly with me	<input type="radio"/>				
2. People call me names.	<input type="radio"/>				
3. People avoid looking at me	<input type="radio"/>				
4. People I don't know act surprised or startled when they see me	<input type="radio"/>				
5. People are nice to me	<input type="radio"/>				
6. People don't know what to say to me	<input type="radio"/>				
7. People I don't know say "Hi" to me	<input type="radio"/>				
8. People laugh at me	<input type="radio"/>				
9. People are relaxed around me	<input type="radio"/>				
10. People feel sorry for me	<input type="radio"/>				
11. People pick on me	<input type="radio"/>				

12. People I don't know smile at me in a friendly way.	<input type="radio"/>				
13. People don't know how to act around me	<input type="radio"/>				
14. People do "double takes" or turn around to look at me	<input type="radio"/>				
15. People are kind to me	<input type="radio"/>				
16. People bully me.	<input type="radio"/>				
17. Strangers are polite to me	<input type="radio"/>				
18. People make fun of me	<input type="radio"/>				
19. People I don't know stare at me	<input type="radio"/>				
20. People treat me with respect.	<input type="radio"/>				
21. People seem embarrassed by my looks	<input type="radio"/>				

Now we would like to know how often you feel or think in certain ways. Please say how often you feel or think about the statements below. Using the scale below, please tick **never** to **always**.

**(SOCIAL COMFORT QUESTIONNAIRE)**

1. I feel like I fit in with most groups.	<input type="radio"/>				
2. No one can understand me	<input type="radio"/>				
3. I would rather be by myself than with other people.	<input type="radio"/>				
4. I like meeting new people.	<input type="radio"/>				
5. It is easy for me to talk to other people my age.	<input type="radio"/>				
6. I feel comfortable in a crowd.	<input type="radio"/>				
7. I feel like I don't fit in with other people.	<input type="radio"/>				
8. It is easy for me to blend in with other people.	<input type="radio"/>				

**F: Support and Information**

School: How much do you agree with the following statements? (please tick)

I had/have really good support at school	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
My school didn't/doesn't really understand NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
NF1 didn't make any difference at school	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Any other comments about school?

Support: Some people tell us they sometimes feel down about NF1, if you ever feel down what helps you feel better? (Please tick all those that help)

- Friends
- Family
- Boyfriend/girlfriend
- Online support groups
- Helplines
- Support groups
- I don't feel down
- other (please explain) \_\_\_\_\_

Is there any support you think should be available for young people with NF1?

Where do or did you find information about NF1? ( Please tick all that apply)

- I've never really looked for information
- Medical Staff
- Family
- Friends
- internet
- the Neuro Foundation/NF Association
- other \_\_\_\_\_

Any comments about your experiences of getting information about NF1?

Do you agree or disagree with the following statements? (please tick)

Most people have heard of NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
TV, magazines and the internet show realistic examples of NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The internet is a good place to find information on NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Having NF1 would be easier if more people knew about it and understood it	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Would you like to find out more about the topics in this survey? Please find below some of useful links:

Centre for Appearance Research ([www.hls.uwe.ac.uk/research/car.aspx](http://www.hls.uwe.ac.uk/research/car.aspx))

A centre for psychological and interdisciplinary research in appearance, disfigurement, body image and related studies.

Neuro Foundation ([www.nfauk.org](http://www.nfauk.org))

A UK charity that supports people with Neurofibromatosis

Changing Faces ([www.changingfaces.org.uk](http://www.changingfaces.org.uk))

A UK charity which supports anyone with a visible difference.

Would you like Feedback on the findings of this survey? Please leave your email/postal address if you would be interested in a copy of the findings of this survey

Would you like to be entered into the prize draw for a chance to win a £25 High Street or online voucher? To be entered into the prize draw please write your email address or name and address below:

Want to contact the researchers?

Please call the researcher Jenny Barke on 0117 3281891 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

Postal address: Jenny Barke, Centre for Appearance Research, Department of Psychology, University of the West of England, Coldharbour Lane, Bristol, BS16 1QY

Supervisor: Dr Diana Harcourt, Reader in Health Psychology and co-director of the Centre for Appearance Research at the University of the West of England

Email: [diana2.harcourt@uwe.ac.uk](mailto:diana2.harcourt@uwe.ac.uk)

**Thank you for taking part**

**The online version of the survey included the question below which replaced the consent section of the above survey:**

If you are under 16 please type in below your parent/carers email or postal address so we can contact them and ask for their permission

**The email below was then sent to parents:**

Dear Parent/Carer

I am contacting you as your child has completed a questionnaire in a research project that is investigating young peoples' experiences of NF1. As your child is under 16 we would like your consent to include their responses in our research.

Please find attached an information sheet and example questionnaire.

If you are happy for your child to take part we would be grateful if you could reply to this email saying:

**I agree to my child taking part in the study**

For further information please contact me, Jenny Barke, on 0117 3281891 or by return email. Also if you would like to take part in the research there is a survey for parents of young people who have NF1 which can be found at: [www.tinyurl.com/NF1parents](http://www.tinyurl.com/NF1parents)

Thank you

Jenny Barke

Centre for Appearance Research

University of the West of England, Department of Psychology, Coldharbour Lane, Bristol, BS16 1QY

### Appendix 19: From themes to survey questions: Parents

Theme	Key aspect	Main points to take forward	Questions/measures on questionnaire
Information and Support	Perceptions of NF1	How did parents manage their child's initial diagnosis of NF1? What were parent's main concerns and have these changed over time?	<p><i>"Have you told family about the diagnosis?" / "Did the diagnosis impact on decision to have further children?"</i></p> <p><i>"When my child was first diagnosed with NF1 my main concern was...." and "Now my main concern is..."</i></p>
		How do people interact with health professionals?	Direct questions about healthcare using quotes from interviews and asking if respondents agree/disagree
	Coping Strategies	How did school support their child's educational needs?	Direct questions about school support using quotes from interviews and asking if respondents agree/disagree
		How do parents seek information and support?	Direct questions regarding information and support including <i>"Where did/do you find information and support?"</i>
Challenge	Perceptions of NF1	How/is their child affected by NF1 and what support would be useful?	<p>Brief IPQ (questions about child)</p> <p><i>"Could you briefly describe the main ways in which your child's NF1 affects them?"</i></p> <p><i>"What support would you like to see available for young people with NF1?"</i></p>
		How do people think having a child with NF1 has impacted on their life?	<p><i>"Could you briefly describe the main ways in which your child's NF1 affects you?"</i></p> <p>Brief IPQ (questions about parent)</p>

	<b>Coping Strategies</b>	<p>What strategies are used to manage any impact and what support and information has been useful?</p> <p>How do media portrayals and general knowledge of NF1 impact on their experience?</p>	<p>Coping Health Inventory for Parents (CHIP)</p> <p><i>Where do you look for information and support? Any other comments? Plus open ended question-What support would you like to see available for parents of children with NF1?</i></p> <p>Questions using quotes from interviews and asking if respondents agree/disagree</p>
<b>Appearance</b>	<b>Appearance &amp; Noticeability</b>	<p>How important do parents think appearance concerns are to their children and are they confident managing any concerns?</p>	<p><i>How often does your child express concern about (a) their appearance in general? (b) Their appearance related to NF1?</i></p> <p><i>How confident are you in managing any concerns? (page 9)/Has your child's attitude to their appearance changed at any point? (if so why)</i></p>
		<p>How noticeable do parents feel their child's NF1 is? How/does this relate to their perceptions of the impact of NF1 on their child?</p>	<p>Parents asked to rate how noticeable their child's NF1 is.</p>
	<b>Social situations</b>	<p>How do others react to their child and how does their child manage reactions from others?</p>	<p>PSQ and SCQ (perceived stigma and social comfort)</p>

## Appendix 20: Information sheets and survey for parents

**Measures used (B-IPQ, CHIP PSQ and SCQ) are indicated in capital letters and underlined for the purpose of this appendix-this did not appear on surveys used in the study.**

You are invited to take part in a research study. Before you decide whether or not you would like to take part it is important that you know why the research is being carried out and what you will be asked to do. This sheet aims to give you the information you need so that you can decide if this study is something you would like to take part in. If there are any points you are not clear on please do ask questions. Thank you for reading this sheet.

### What is the purpose of the study?

We would like to understand how young people experience having NF1. There has not until now been much research that focuses on how NF1 impacts specifically on young people. If we can understand how young people with NF1 feel and how they manage their condition then we can try to help to make things better for other young people and their families in the future.

This study is being carried out by researchers at the Centre for Appearance Research at the University of the West of England in Bristol. This study will be part of a research degree (PhD) for one of the researchers, Jenny Barke.

We would like to ask you to think about how having NF1 may or may not affect your child. Your child might also be deciding whether or not to take part in the study (please see the young person's information sheet) and you may wish to discuss this with them.

### Who can take part in the study and do I have to?

We would like young people aged 14-24 who have a diagnosis of NF1, and their parents/carers to take part in this research. You do not have to take part. It is totally up to you and if your child is under 16 we would also need your permission for them to take part.

If you do decide to take part completing the survey will be taken as consent to participate. If your child is under 16 and wants to take part we would also need your signed consent on their questionnaire. If you or your child change your mind about taking part you can stop at any time; no one will mind or be upset.

You and your child do not have to both take part, surveys can be completed by you, your child or you can both take part.

### What will I have to do?

If you decide you would like to take part we would like you to complete the parent questionnaire enclosed, it is about your experience so there are no right or wrong answers.

We understand that some aspects of this topic may be personal and sensitive; it's fine if you don't want to answer any questions and if you feel uncomfortable you can stop at any time.

There is also an online version of the questionnaire which can be found at **parents link** please feel free to complete either version.

### What are the benefits?

We don't expect there will be any direct benefits for you; some people find it interesting to take part in research and to think about their experiences. The information will help us to understand your

experiences and we hope that overall this research programme will help to inform the provision of care and support for young people with NF1.

**What happens to the results and will my taking part be kept private?**

The results of this study will be shared with health professionals, charities, support groups and people involved in research in this area. It may be reported in professional journals or at meetings and conferences.

However you will not be identified or mentioned by name at any point. Your responses will be kept safe and treated as confidential and we will not discuss what you tell us with anyone else.

**Who is supporting this study?**

All research in the NHS is looked at by an independent group of people, called a Research Ethics Committee. Research Ethics Committees (RECs) safeguard the rights, safety, dignity and well-being of people participating in research in the National Health Service. They review applications for research and give an opinion about the proposed participant involvement and whether the research is ethical. This study has been reviewed and given favourable opinion by NRES Committee South West – Frenchay.

**What next?**

Thank you for taking the time to read this. If you are interested in taking part or would like to ask any questions please call the researcher, whose name is Jenny Barke, on 07500121137 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

As a thank you for your time completing the survey you can choose to be entered into a prize draws with a chance to win £25 worth of Amazon or High Street shopping vouchers. If you would like to be entered please return your email address in the envelope marked 'parent'.

Jenny Barke, PhD Researcher, Centre for Appearance Research, University of the West of England.

Supervisor: Dr Diana Harcourt, Reader in Health Psychology and co director of the Centre for Appearance Research at the University of the West of England. Email: [Diana2.Harcourt@uwe.ac.uk](mailto:Diana2.Harcourt@uwe.ac.uk)  
Tel: 0117 3283967

**Young Peoples' experiences of NF1**

**Parents Experiences**

**You are invited to take part in a research study. Before you decide whether you want to complete this questionnaire. Please read the information sheet. If you have any questions please contact the researcher whose name is Jenny Barke on 0117 3281891 or email [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk)**

**Please note completing the survey will be taken as consent to participate**

**Please return the completed questionnaire in the envelope provided**

**CREATE YOUR OWN ID CODE**

You have the right to withdraw from the study up to four weeks after you have completed the survey.

Should you wish to withdraw, you will need to inform us by emailing [Jenny.barke@uwe.ac.uk](mailto:Jenny.barke@uwe.ac.uk) quoting your unique ID code. This will enable us to identify all the material that needs to be deleted to withdraw you from the project.

**To create your ID code please enter the first three letters of your first name and the date (day of the month) you were born.**

*If your first name is Karen and you were born on the 5<sup>th</sup> June you would enter **KAR05***

**Please enter your code in the box below:**

**A: About you**

Are you

- Male
- Female

How would you describe your ethnic group?

White

- English / Welsh / Scottish / Northern Irish / British
- Irish
- Gypsy or Irish Traveller
- Any other White background please write in \_\_\_\_\_

Mixed / multiple ethnic groups

- White and Black Caribbean
- White and Black African
- White and Asian
- Any other Mixed / multiple ethnic background please write in \_\_\_\_\_

Asian / Asian British

- Indian
- Pakistani
- Bangladeshi
- Chinese
- Any other Asian background, please write in \_\_\_\_\_

Black / African / Caribbean / Black British

- African
- Caribbean
- Any other Black / African / Caribbean background, please write in \_\_\_\_\_

Other ethnic group

- Arab
- Any other ethnic group, please write in \_\_\_\_\_

Or

- prefer not to answer

Which area do you live in?

- England
- Scotland
- Wales
- Northern Ireland
- Ireland
- Outside UK and Ireland please say which country below \_\_\_\_\_

Have you been given a diagnosis of Neurofibromatosis Type 1 (NF1)

- Yes
- No

Do you have a child aged 14-24 with NF1?

(If you have more than 1 child aged 14-24 with NF1 could you complete this survey with 1 particular child in mind)

- Yes
- No

How old are they?

- 14
- 15
- 16
- 17
- 18
- 19
- 20
- 21
- 22
- 23
- 24

Are they

- male
- Female

Is their NF1 inherited or a new diagnosis?

- Inherited
- New
- Not sure

Roughly how old were they when they were first diagnosed?

- under 1
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10
- 11
- 12

- 13
- 14
- 15
- 16
- 17
- 18
- 19
- 20
- 21
- 22
- 23
- 24

Does anyone else in your family have NF1?

- Yes -if yes please explain what relation they are to you \_\_\_\_\_
- don't know
- No

Have you told any members of your family about your son/daughters diagnosis?

- Yes
- No

If no, could you briefly explain why not?

Did the initial diagnosis of your child impact on your decisions regarding having any further children?

- yes
- No

Could you explain how?

What health professionals does your son/daughter see for NF1?

- GP
- Genetics specialist
- NF1 Specialist
- Children's doctor or nurse
- Dermatologist (skin)
- Eye Specialist
- Don't know
- Other (please explain) \_\_\_\_\_

Could you briefly describe the main ways in which your child's NF1 affects you

Could you briefly describe the main ways in which your child's NF1 affects them

**B: Managing NF1**

Please complete the following sentences:

When my child was first diagnosed with NF1 my main concern was....

Now my main concern is.....

**(COPING HEALTH INVENTORY FOR PARENTS)**

Nf1 affects people and families in different ways and people experience and cope with having a child with NF1 in different ways. In this section you are asked to read the list of different behaviours below and for each behaviour you have used please tick how helpful it was:

extremely helpful      moderately helpful      minimally helpful      not helpful

For each behaviour you did not use please show your reason by ticking in one of the 2 boxes:

Chose not to use it    or    Not possible

Please read and record your decision for each behaviour listed below.

	extremely helpful	moderately helpful	minimally helpful	not helpful	I do not cope this way because chose not to	not possible
Talking over personal feelings and concerns with partner.	<input type="radio"/>	<input type="radio"/>				
Engaging in relationships and friendships which help me to feel important and appreciated	<input type="radio"/>	<input type="radio"/>				
Trusting my partner (or former partner) to help support me and my child(ren).	<input type="radio"/>	<input type="radio"/>				
Sleeping	<input type="radio"/>	<input type="radio"/>				
Talking with the	<input type="radio"/>	<input type="radio"/>				

<p>medical staff (nurses, social worker, etc.) when we visit the hospital/clinic.</p>						
<p>Believing that my child/ren will get better</p>	<input type="radio"/>					
<p>Working, employment</p>	<input type="radio"/>					
<p>Showing that I am strong</p>	<input type="radio"/>					
<p>Buying gifts for myself and/or other family members</p>	<input type="radio"/>					
<p>Talking with other people/parents in my same situation</p>	<input type="radio"/>					
<p>Taking good care of all the medical equipment at home</p>	<input type="radio"/>					
<p>Eating</p>	<input type="radio"/>					
<p>Getting other members of the family to help with chores and tasks at home</p>	<input type="radio"/>					
<p>Getting away by myself</p>	<input type="radio"/>					
<p>Talking with the doctor about my concerns about my child/ren with NF1</p>	<input type="radio"/>					
<p>Believing that the GP/hospital has</p>	<input type="radio"/>					

my family's best interest in mind						
Building close relationships with people	<input type="radio"/>					
Believing in God	<input type="radio"/>					
Develop myself as a person	<input type="radio"/>					
Talking with other parents in the same type of situation and learning about their experiences	<input type="radio"/>					
Doing things together as a family (involving all members of the family)	<input type="radio"/>					
Investing time and energy in my job	<input type="radio"/>					
Believing that my child is getting the best medical care possible	<input type="radio"/>					
Entertaining friends in our home	<input type="radio"/>					
Reading about how other people in my situation handle things	<input type="radio"/>					
Doing things with family relatives	<input type="radio"/>					
Becoming more self reliant and	<input type="radio"/>					

independent						
Telling myself I have many things I should be thankful for	<input type="radio"/>					
Concentrating on hobbies (art, music, jogging etc)	<input type="radio"/>					
Explaining family situation to friends and neighbours so they will understand us	<input type="radio"/>					
Encouraging child/ren with NF1 to be more independent	<input type="radio"/>					
Keeping myself in shape and well groomed	<input type="radio"/>					
Involvement in social activities (parties etc) with friends	<input type="radio"/>					
Going out with my partner on a regular basis	<input type="radio"/>					
Being sure prescribed medical treatments for my chid/ren are carried out at home on a daily basis	<input type="radio"/>					
Building a closer relationship with my partner	<input type="radio"/>					

Allowing myself to get angry	<input type="radio"/>					
Investing myself in my child/ren	<input type="radio"/>					
Talking to someone (not professional counsellor/doctor) about how I feel	<input type="radio"/>					
Reading more about NF1	<input type="radio"/>					
Trying to keep family stability	<input type="radio"/>					
Being able to get away from the home care tasks and responsibilities for some relief	<input type="radio"/>					
Having my child with NF1 seen at the GP's/hospital on a regular basis	<input type="radio"/>					
Believing that things will always work out	<input type="radio"/>					
Doing things with my children	<input type="radio"/>					

Where do or did you find information and support related to NF1? (Please tick all that apply):

- I've never really looked for information
- Medical Staff
- Family
- internet
- the Neuro Foundation/NF Association
- NF Specialist advisor
- Friends
- Partner
- Online support groups
- Helplines
- Support groups (not online)
- other (please explain) \_\_\_\_\_

Any comments on information and support?

**C: My child and NF1 (BRIEF IPQ-ADAPTED)**

For the following questions please circle the number that matches what you think

How much does NF1 affect your child's life?

0	1	2	3	4	5	6	7	8	9	10
No affect at all									severely affects their life	

How much does NF1 affect your life?

0	1	2	3	4	5	6	7	8	9	10
No affect at all									severely affects my life	

How long do you think your child's NF1 will continue?

0	1	2	3	4	5	6	7	8	9	10
A very short time									Forever	

How much control do you feel you have over your child's NF1?

0	1	2	3	4	5	6	7	8	9	10
Absolutely										extreme amount
no control										of control

How much do you think treatment can help NF1?

0	1	2	3	4	5	6	7	8	9	10
Not at all										Extremely helpful

How much does your child experience symptoms from NF1?

0	1	2	3	4	5	6	7	8	9	10
No symptoms										many severe
at all										symptoms

How concerned are you about your child's NF1?

0	1	2	3	4	5	6	7	8	9	10
Not at all										extremely
concerned										concerned

How well do you feel you understand NF1?

0	1	2	3	4	5	6	7	8	9	10
Don't understand										Understand
at all										very clearly

How well do you feel your child understands NF1?

0	1	2	3	4	5	6	7	8	9	10
Don't understand										Understand
at all										very clearly

How much does NF1 affect you emotionally (eg does it make you angry, scared, upset or depressed?)

0      1      2      3      4      5      6      7      8      9      10

Not at all extremely affected

affected emotionally emotionally

How much does NF1 affect your child emotionally (eg does it make them angry, scared, upset or depressed?)

0      1      2      3      4      5      6      7      8      9      10

Not at all extremely affected

affected emotionally emotionally

Please list (most important first) the 3 most important things that you believe caused your child's NF1. The most important causes for me:-

- 1
- 2
- 3

**D: Support and understanding:** Do you agree or disagree with the following statements? (please tick)

	Strongly Agree	Agree	Disagree	Strongly Disagree
My child had/has good support at school	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
My child's school didn't/doesn't understand NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
It was a battle to get the right support for my child at school	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
NF1 didn't make any difference to my child at school	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Medical staff generally know about and understand NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

I sometimes have to explain to medical staff what NF1 is	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Most medical staff don't know about NF1, I have to be the expert	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Most people have normally heard of NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I'm happy to explain NF1 to other people	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
TV magazines and the internet show realistic examples of NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The internet is a good place to find information on NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Having NF1 would be easier if more people knew about it and understood it	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Charities and support groups show positive images of people with NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

**Section E: Appearance**

E1 How noticeable is your child's NF1 to other people on a day to day basis?

0 1 2 3 4 5 6 7 8 9 10

Not at all

Highly

Noticeable

Noticeable

How often does your child express concern about their appearance in general?

- Daily
- Sometimes
- Rarely
- Never



**(PERCEIVED STIGMATIZATION QUESTIONNAIRE)**

During your child's normal day, they probably see and talk to many different people. Based on your observations over the last year, please rate how often people act in certain ways toward your child and how they react emotionally. Please give your opinion. Do not ask your child.

First, for each question, tick how often people do certain things using the scale below, never to always

1. People are friendly with my child	<input type="radio"/>				
2. People call my child names.	<input type="radio"/>				
3. People avoid looking at my child	<input type="radio"/>				
4. People who don't know my child act surprised or startled when they see him/her	<input type="radio"/>				
5. People are nice to my child	<input type="radio"/>				
6. People don't know what to say to my child	<input type="radio"/>				
7. People my child doesn't know say "Hi" to him/her	<input type="radio"/>				
8. People laugh at my child	<input type="radio"/>				
9. People are relaxed around my child	<input type="radio"/>				
10. People feel sorry for my	<input type="radio"/>				

child					
11. People pick on my child	<input type="radio"/>				
12. People my child doesn't know smile at him/her in a friendly way.	<input type="radio"/>				
13. People don't know how to act around my child	<input type="radio"/>				
14. People do "double takes" or turn around to look at my child	<input type="radio"/>				
15. People are kind to my child	<input type="radio"/>				
16. People bully my child	<input type="radio"/>				
17. Strangers are polite to my child	<input type="radio"/>				
18. People make fun of my child	<input type="radio"/>				
19. People my child doesn't know stare at them	<input type="radio"/>				
20. People treat my child with respect.	<input type="radio"/>				
21. People seem embarrassed by my child's looks	<input type="radio"/>				

**(SOCIAL COMFORT QUESTIONNAIRE)**

Now we would like to know how often you think your child feels or thinks in certain ways. Please say how often you think your child feels or thinks the statements below. We are interested in your opinion-please do not ask your child.

	Never	Almost never	Sometimes	Often	Always
He/she feels like they fit in with most groups	<input type="radio"/>				
He/she feels that no one understands them	<input type="radio"/>				
He/she would rather be by themselves than with other people	<input type="radio"/>				
He/she likes meeting new people	<input type="radio"/>				
It is easy for him/her to meet other people his/her own age	<input type="radio"/>				
He/she feels comfortable in a crowd	<input type="radio"/>				
He/she feels like he/she does not fit in with other people	<input type="radio"/>				
It is easy for him/her to blend in with other people	<input type="radio"/>				

What support, if any, would you like to see available for parents of children with NF1?

What support, if any, would you like to see available for young people with NF1?

Would you like to find out more about the topics in this survey?

Please find below some of useful links:

Centre for Appearance Research ([www.hls.uwe.ac.uk/research/car.aspx](http://www.hls.uwe.ac.uk/research/car.aspx))

A centre for psychological and interdisciplinary research in appearance, disfigurement, body image and related studies.

Neuro Foundation ([www.nfauk.org](http://www.nfauk.org))

A UK charity that supports people with Neurofibromatosis

Changing Faces ([www.changingfaces.org.uk](http://www.changingfaces.org.uk))

A UK charity which supports anyone with a visible difference.

Would you like Feedback on the findings of this survey? Please leave your email/postal address if you would be interested in a copy of the findings of this survey

Would you like to be entered into the prize draw for a chance to win a £25 High Street or online shopping voucher? To be entered into the prize draw please write your email address or name and address below:

Want to contact the researchers?

Please call the researcher Jenny Barke on 0117 3281891 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

Postal address: Jenny Barke, Centre for Appearance Research, Department of Psychology, University of the West of England, Coldharbour Lane, Bristol, BS16 1QY

Supervisor: Dr Diana Harcourt, Reader in Health Psychology and co-director of the Centre for Appearance Research at the University of the West of England, Email: [diana2.harcourt@uwe.ac.uk](mailto:diana2.harcourt@uwe.ac.uk)

**Thank you for taking part**

### Appendix 21: From themes to survey questions health professionals

Themes	Key aspect	Main points to take forward	Questions on questionnaire
The role of the professional	Engagement with Healthcare and Support	<p>Is being a point of contact a large part of professionals roles?</p> <p>How often do professionals feel their role is related to explaining a diagnosis and putting information into context?</p> <p>Do professionals feel their role includes supporting reproductive decision making and providing psychosocial support?</p>	<p><i>Direct question asking for professionals role and contact with patients with NF1.</i></p> <p><i>Question states 'my role involves....' and respondents are asked to identify how often their role involves a series of different activities (as identified in interviews).</i></p> <p><i>Respondents are also asked whether they agree or disagree with a series of statements including some related to their role and contact with patients</i></p>
		<p>Is supporting families and parents an important aspect of professional's role?</p> <p>Do parents want advice support and information on talking to their children?</p>	<p><i>Respondents are also asked whether they agree or disagree with a series of statements including some related to their different relationships within families</i></p>
Family	Inheritance	<p>Do professionals feel that families differ in their management of NF1 if it is inherited or De novo?</p>	<p><i>Respondents are asked whether they agree or disagree with a series of statements including some related to the differences between families where NF1 is a new or pre existing condition.</i></p> <p><i>Plus open ended questions:</i></p> <p><i>(a) The most common concern for families where the diagnosis is new is.....</i></p> <p><i>(b) The most common concern for families where the diagnosis is a pre existing condition is.....</i></p>
Adolescence	Engagement with Healthcare	<p>Do professionals see young people on a regular basis?</p> <p>Is it difficult to reach young people?</p>	<p><i>Asked to agree/disagree with a series of statements on regularity/type of contact with young people</i></p> <p><i>Asked to agree/disagree 'it's difficult to reach young people in practical terms'</i></p>

Appearance	Appearance concerns and support	Do professionals feel it is 'normal' for appearance to become more of a concern in adolescence for young people generally and young people with NF1 in particular?	<p>Asked to agree/disagree with a series of statements about appearance.</p> <p>If you agreed appearance concerns change during adolescence why is this? (series of answers plus text box for more detail/'other')</p>
		What strategies do health professionals feel people employ to manage appearance concern?	Asked to agree/disagree with a series of statements about appearance.
		<p>Do young people report concerns? What concerns do professionals feel young people may have?</p> <p>What support is needed to support young people and their families?</p>	<p>The most common concern for young people is.....</p> <p>Asked to agree/disagree with statements regarding support (ease of access, types of support and support groups)</p> <p>Support I would like to see developed would be....(tick boxes plus section for 'other'.)</p>

## **Appendix 22: Information sheets and survey for health professionals**

**You are being invited to take part in this study. Before you decide if you would like to participate, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and email the researcher if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.**

### **Who is carrying out the research?**

This project is being carried out by researchers at the Centre for Appearance Research at the University of the West of England. This study will be part of a research degree (PhD) for one of the researchers, Jenny Barke.

### **What is the purpose of the study?**

Despite the potential impact of NF1 on quality of life and psychosocial adjustment during adolescence there is limited research that examines the actual lived experience of young people with NF1. Therefore this research aims to explore the psychosocial impact of NF1 on adolescents' lived experience by examining both resilience and the challenges being faced, in order to identify any support needs and the factors that might help positive adjustment. The study you are invited to take part in is a survey for healthcare specialists who work with young people with genetic conditions including NF1. This is being undertaken in order to get an insight into current provision and care for young people with NF1 and possible support needs.

### **What will participation involve and how long will it take?**

If you agree to take part in the study you will be required to fill in an online or paper survey. The survey will take approximately 10 minutes of your time and will require you answering a number of questions on a range of topics about your work with families and young people with NF1.

### **What about confidentiality?**

The information you give us will be treated with the highest level of confidentiality. You will be prompted to generate a unique participant identification code. Information that would make it possible to identify you or any other participant will never be included in any sort of report.

Your responses will be written up and the data may be published in an academic journal or elsewhere, and although direct quotes from you may be used in a paper or report, your name and identifying information will be kept anonymous. The data will only be accessible to those working on the project.

### **Do I have to complete the whole survey?**

Your participation in this research is entirely voluntary. You have the right to answer as many or as few of the questions asked as you wish. If you decide you want to withdraw from the study after you have finished the survey, you can email us with your participant ID code (which you will generate at the start of the survey) up to 4 weeks after completing the study, so that we can identify and delete your responses.

### **What are the possible disadvantages and risks of taking part?**

Any participation in research can raise sensitive issues or painful emotions but also positive insights. It is entirely your choice as to what you want to share with the researchers via the survey. We would also like to reassure you that there is no right or wrong answer and that no judgments will be made on the basis of what you write.

**What are the possible benefits of taking part?**

We hope you will find participation in this project insightful and interesting. We hope that overall this research programme will help to inform the provision of care and support for young people. In addition if you leave your email address you will be entered into a prize draw for the chance to win a £50 M&S voucher.

**Who is supporting this study?**

This study has been approved by the Ethics Committee at the Faculty of Health and life Sciences at The University of the West of England.

**Do you have any questions about the research?**

Thank you for taking the time to read this. If you are interested in taking part or would like to ask any questions please call the researcher, Jenny Barke, on 0117 3281891 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

**All participants are eligible for entry into a prize draw for a £50 M&S voucher, to be entered please give your email address at the end of the survey**

Jenny Barke, PhD Researcher Centre for Appearance Research,  
University of the West of England.

Dr Diana Harcourt, Reader in Health Psychology and co director of the Centre for Appearance Research, University of the West of England

Prof Jane Coad, Professor in Children and Family Nursing, Coventry University

Dr Ainsley Newson, Senior Lecturer in Biomedical Ethics at the Centre for Ethics in Medicine at Bristol University.

**Thank you for showing an interest in this project**

**Before you decide if you would like to participate in this survey, it is important for you to understand why the research is being done and what it will involve. Please take time to read the information sheet and if you have any questions please contact the researcher, Jenny Barke on 0117 3281891 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).**

**Once you have completed the survey please return it in the pre paid envelope provided. This survey is also available online: [http://uwepsych.qualtrics.com/SE/?SID=SV\\_5cDWazYawil5bGA](http://uwepsych.qualtrics.com/SE/?SID=SV_5cDWazYawil5bGA)**

**Please note completing this survey is taken to be consent that you  
are happy to take part in this study**

**Jenny Barke, Centre for Appearance Research, University of the West of England.**

**Supervisor; Dr Diana Harcourt, Reader in Health Psychology and co director of the Centre for Appearance Research at the University of the West of England.**

**Email: [Diana2.Harcourt@uwe.ac.uk](mailto:Diana2.Harcourt@uwe.ac.uk) Tel: 0117 3283967**

#### **CREATE YOUR UNIQUE PARTICIPANT IDENTIFICATION CODE**

You have the right to withdraw from the study up to four weeks after you have completed the survey. Should you wish to withdraw, you will need to inform us by emailing [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk) quoting your unique participant identification code. This will enable us to identify all the material that needs to be deleted due to your withdrawal from the project.

To create your unique participant identification code please enter the first three letters of your mother's maiden name and the date (day of the month) you were born.

If your mother's maiden name is Smith and you were born on the 23rd June you would enter SMI23 Please enter your code in the box below.

**My job title is.....**

- Nurse/Specialist Nurse (Please indicate specialism) \_\_\_\_\_
- Paediatrician
- Geneticist
- Psychologist
- Social worker
- Genetic Counsellor
- Consultant Surgeon
- other \_\_\_\_\_

**I see patients with Neurofibromatosis Type 1.....**

- Never
- Once a Year or Less
- Several Times a Year
- Once a Month
- 2-3 Times a Month
- Once a Week
- 2-3 Times a Week
- Daily

**My role involves.....**

	Never	Rarely	Occasionally	Often	Very Often
Explaining the genetics of the condition to patients and families and supporting initial diagnosis	<input type="radio"/>				
Supporting young people and families to manage the practical side of the condition	<input type="radio"/>				
Monitoring and treating the medical and/or surgical aspects of NF1	<input type="radio"/>				
Signposting young people and families towards support groups and avenues of support	<input type="radio"/>				
Being a point of contact	<input type="radio"/>				
Putting information in context for patients and families	<input type="radio"/>				
Providing psychosocial support	<input type="radio"/>				
Supporting reproductive decision making	<input type="radio"/>				
Other: <div style="border: 1px solid black; height: 15px; width: 250px; margin-top: 5px;"></div>	<input type="radio"/>				

**Please indicate whether you agree/disagree with the following statements:**

	Strongly Disagree	Disagree	Neither Agree nor Disagree	Agree	Strongly Agree
I see patients on a regular basis for routine appointments	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
If patients have concerns related to NF1 they can call and I'll see them anytime	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I worry that people might not contact me or know who to contact if there is a concern related to their condition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The internet is a useful way for patients to find information about their condition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Patients shouldn't Google their condition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
An important part of my role is to support families in talking to their children about NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Parents are best placed to give children information about their condition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I worry sometimes that parents haven't fully informed their children about NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The way people manage their condition depends on who else in the family has NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The concerns that parents have depends on who else in the family has NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Parents are often concerned about how they will talk to their children about NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

**The most common concern for families where the diagnosis of NF1 is new to the family is:**

**The most common concern for families where NF1 is a pre existing condition is:**

**Please indicate if you agree/disagree with the following statements:**

	Strongly Disagree	Disagree	Neither Agree nor Disagree	Agree	Strongly Agree
I tend to see people with NF1 at a younger age or when they're adults thinking about having families of their own	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I see young people regularly	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Very few young people will actively manage their healthcare	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
It's important to allow the family to decide how involved young people are in their care	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
It's difficult to reach young people, in practical terms	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Transition from pediatric to adult services is a time I might see young people	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

**The most common concern for young people with NF1 is:**

Please indicate if you agree/disagree with the following statements:

	Strongly Disagree	Disagree	Neither Agree nor Disagree	Agree	Strongly Agree
Young children with NF1 are normally fairly positive about their appearance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Adolescence is a time people start to worry about their appearance in general	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Most young people with NF1 are concerned about how they look	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
People with NF1 change how they view their appearance between childhood and adulthood	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Its hard for young people with NF1 to talk about appearance concerns	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Young people with NF1 worry about neurofibromas appearing	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The uncertainty of NF1 is the biggest concern for many young people who have the condition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Tumor load is not an indicator of how an individual with NF1 feels about their appearance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Parents are often concerned about how to prepare their children for possible changes in appearance related to NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
It is difficult for all young people to talk about their appearance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

**If you agreed that appearance concerns change during adolescence, why do you think this is?**

	Yes	Maybe	No
Its a normal part of being a young person	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The influence of the media	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The influence of friends	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Neurofibromas appearing	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Celebrity culture	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Interest in dating	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Other, please specify:

**Please indicate if you agree/disagree with the following statements:**

	Strongly Disagree	Disagree	Neither Agree nor Disagree	Agree	Strongly Agree
My service has links with a psychological service/therapeutic counselling	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
If a patient needed ongoing psychological support there is a clear pathway	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
It's difficult to access psychological services	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Patients are often involved in support groups	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Patients don't want to be part of a support group	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I don't think people with NF1 identify with any particular support group	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I think people with NF1 prefer to deal with their condition within their family	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Families are biggest source of support for young people with NF1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
I think young people with NF1 need easy access to more support services	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

**Support I would like to see developed for young people with NF1 would be:**

	Yes	Maybe	No
Local support groups for young people	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Online support and forums for young people	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Transition groups	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Transition programme	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Information specific for young people	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Summer camps/away days	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Helplines	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Supporting families early on and preparing young people	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Other	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

**If other please explain:**

**Would you like to find out more about the topics in this survey?**

Please find below a number of useful links from medical information to support groups/charities set up to help people affected by different body or appearance-related conditions.

**Centre for Appearance Research** ([www.hls.uwe.ac.uk/research/car.aspx](http://www.hls.uwe.ac.uk/research/car.aspx)) A centre for psychological and interdisciplinary research in appearance, disfigurement, body image and related studies.

**Neuro Foundation** ([www.nfauk.org](http://www.nfauk.org)) A UK charity that supports people with Neurofibromatosis

**Changing Faces** ([www.changingfaces.org.uk](http://www.changingfaces.org.uk)) A UK charity which supports anyone with a disfigurement.

**Would you like to be further involved in this research?**

We would welcome your help in identifying and recruiting young people aged 11-18 and their parents to take part in a survey. If you are interested in this or in receiving a report on the findings of this research please leave your email details below. Alternatively findings will be available on the Centre for Appearance Research website from September.

**Would you like to be entered into a prize draw to win a £50 M&S voucher?**

If so please leave your email address below, all entries need to be received by the 30th June 2011 and the winner will be contacted by the 8th July 2011

**Want to contact the researchers?**

Please call the researcher, Jenny Barke, on 0117 3281891 or you can email her at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk).

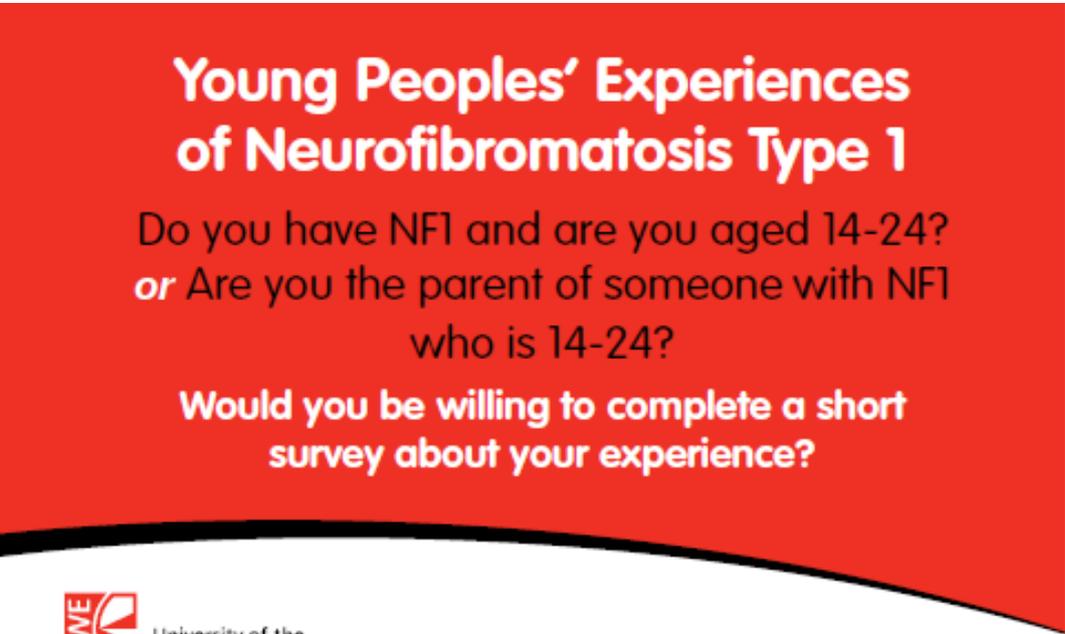
Postal address:

Jenny Barke, Centre for Appearance Research, Department of Psychology, University of the West of England, Coldharbour Lane, Bristol, BS16 1QY

## Thank you for your participation

## Appendix 23: Recruitment adverts for surveys of young people and parents

Recruitment information placed online for young people and parents:



**Young Peoples' Experiences  
of Neurofibromatosis Type 1**

Do you have NF1 and are you aged 14-24?  
*or* Are you the parent of someone with NF1  
who is 14-24?

**Would you be willing to complete a short  
survey about your experience?**

 University of the  
West of England

**bettertogether**



**We want to hear about your experience of NF1.  
Please take part in this brief online survey to share your  
views.**

As a thank you for taking part in this study you can choose to be  
entered into a draw to win **one of 2 £25 shopping vouchers.**

For more information and to access the online survey please  
use one of the following links:

[www.tinyurl.com/NF1parents](http://www.tinyurl.com/NF1parents)  
[www.tinyurl.com/NF1yp](http://www.tinyurl.com/NF1yp)

Or if you would prefer a paper copy please contact Jenny with  
your address.

Thank you very much

**Jenny Barke, Centre for Appearance Research**  
University of the West of England,  
Coldharbour Lane, Bristol BS16 1QY  
**E-mail** [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk) **Text** 07500 121137

## **Appendix 24 : Recruitment information from The Neuro Foundation Newsletter regarding surveys of young people and parents**

Hello my name is Jenny; I am a researcher at the Centre for Appearance Research, at the University of the West of England in Bristol. I am working on a research degree (PhD) investigating young peoples' experience of NF1. The aim of this research is to explore young people's experience of NF1. Past research has suggested that adolescence can be a time when having NF1 is particularly difficult for some people. As NF1 is a very variable condition people probably have very different experiences and it would be useful to understand a little more about the challenges people face and also the things people find supportive and helpful. I hope this project as a whole gives people a chance to share their experience and I also hope it will be useful in designing support for young people and their families in the future.

In order to understand young people's experience I started by interviewing young people aged 14-24 with NF1, parents of people aged 14-24 and health professionals who work with people with NF1. Interviews took place in the UK and lasted between half an hour and an hour and a half. Interviews have been analysed by looking for themes across interviews and some of the findings are briefly described below:

Interviews with young people suggest the variability of NF1 was an important issue; it seemed that NF1 meant different things to different people. People had very different concerns including worries about their appearance and many people had either sought support or said they would like support managing these concerns. Some people said it can be difficult to deal with the reactions of other people and answer questions about their appearance and NF1, although others felt it was fairly easy to manage as it was something they were used to. Many young people said they would like NF1 to be better recognised by the general public and also schools and medical staff. Interviews with parents highlighted that people wanted information and support both for themselves and their child. Many parents felt that NF1 affected their whole family in different ways. Some parents had concerns about the affect of NF1 on their child's appearance and felt the visibility of NF1 meant having to deal with questions from other people. Finally, like the young people, parents felt there was a lack of understanding and recognition of NF1 and that it was often misrepresented, especially in the media. Health professionals discussed many of the same concerns as the young people and parents. People felt an important part of their role was in supporting whole families and being able to put information into context. It may be that adolescence is a time when people are not engaged with healthcare for many reasons. Adolescence can be a time when people are particularly worried about the way they look which might bring new challenges to young people who have NF1. An area that was particularly important to people was raising awareness and understanding of NF1, in the general public, in schools and in the medical community. It was also important to many people that information should be readily available for young people and parents to learn about NF1 themselves. People also wanted both social support and practical advice on managing NF1 in terms of their appearance and dealing with uncertainty.

Themes from the interviews have been used to write questionnaires for young people, parents of young people and health professionals to explore issues raised in the interviews in more detail. Questionnaires will be online and on paper from Mid March 2012. For more information or to register your interest in completing a questionnaire please call or text me, Jenny on mobile: 07500 121137 or email [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk) or write to Jenny Barke, Centre for Appearance Research, University of the West of England, Coldharbour Land, Bristol, BS16 1QY

**Appendix 25: Health professionals' survey recruitment information from  
the Newsletter of the British Society for Human Genetics**

## Call for participants: young people's experience of NF1

Jenny Barke, PhD Student, Centre for Appearance Research, University of the West of England, Coldharbour Lane, Bristol, BS16 1QY.

Neurofibromatosis Type 1 (NF1) has been found to have a significant impact on quality of life and psychological adjustment for some individuals (1). However it is impossible to predict what kinds of challenges a person with NF1 may face. Psychological problems can stem from an altered appearance caused by neurofibromas and from the unpredictability of the condition (2). Adolescence is generally recognised as a time during which appearance becomes more significant for young people; it is during this time that neurofibromas often first appear and become particularly noticeable. Not knowing how the condition will progress makes adolescence a time of uncertainty for those with NF1. Despite the potential impact of NF1 on quality of life and psychosocial adjustment during adolescence there is limited research that examines the lived experience of young people with NF1. Therefore this research aims to explore the psychosocial impact of NF1 on adolescents' lived experience by examining both resilience and challenges being faced, in order to identify any support needs and the factors that might help positive adjustment. The overall aim of this research is to inform the future provision of care for young people with NF1.

### **Research plan**

The first stage of this research is a series of exploratory interviews with 1) health professionals; 2) young people aged 14-24 with NF1 and 3) parents of young people with NF1. These interviews will be used to develop surveys to further investigate areas identified in the exploratory interviews. Interviews with young people and parents are currently taking place. If you know of any families or individuals who are suitable for this research and may be interested in taking part in a research interview, please contact Jenny Barke for further information. Interviews with health professionals are complete, and the survey for health professionals who work with people with NF1 can be found at <http://tiny-url.org/NF1>. It takes less than ten minutes to complete and entries can be entered into a draw to win a £50 M&S voucher (Survey closes 30 June 2011). For further information on any aspect of this research please contact Jenny Barke.

### **References:**

1. Graf A, Landolt MA, Mori AC, Boltshauser E, Quality of life and psychological adjustment in children and adolescents with neurofibromatosis type 1. *Journal of Pediatrics* 2006;149:348-353.
2. Ferner RE, Huson SM, Thomas N, Moss C, Willshaw H, Evans DG, Upadhyaya M, Towers R, Gleeson M, Steiger C, Kirby A. Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. *Journal of Medical Genetics (JMG)*, 2007, 44(2), pp. 81.

**Appendix 26: Information for health professionals regarding surveys sent out on emails and e-newsletters/lists**

Dear

I am researching young peoples' experiences of Neurofibromatosis type 1. Within this I am interested in the views of health professionals who work with people with NF1. As a result of a series of interviews with health professionals I have devised a survey which can be found here: <http://tiny-url.org/NF1> it takes less than 10 minutes to complete and gives participants a chance to win a £50 M&S voucher,

For further details or any information please contact me by email ([jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk)) or on 0117 3281891 - thank you in advance.

Best wishes, Jenny Barke  
Centre for Appearance Research, University of the West of England

## Appendix 27: Recruitment letters for survey study sent from clinicians to parents and young people

HOSPITAL/CLINIC ADDRESS

DATE

Dear

I am writing to ask if you would like to take part in a survey which has been designed as part of a research degree by Jenny Barke, a PhD researcher at the University of the West of England, Bristol (UWE). The title of the research is:

Young Peoples' Experience of Neurofibromatosis Type 1

This survey aims to understand the experiences of young people with NF1. If we can understand more about young peoples' experiences of NF1 then we can try to help other young people and their families in the future.

Please find enclosed 2 surveys, one for young people (aged 14-24) and one for parents/carers and further information about the study. If you are interested in taking part please either return the survey in one of the envelopes provided or the survey can be found online. The survey for young people can be found here, [www.tinyurl.com/NF1yp](http://www.tinyurl.com/NF1yp) and the survey for parents can be found here [www.tinyurl.com/NF1parents](http://www.tinyurl.com/NF1parents). As a thank you for your time completing the survey you can choose to be entered into one of 2 prize draws with a chance to win £25 worth of Amazon or High Street shopping vouchers.

Taking part in the study does not involve any extra treatment, follow up or investigations. Taking part is also completely voluntary and if you do decide to take part, you can change your mind at any time. If you choose not to take part at all or if you withdraw at a later stage it will not affect the standard of care you receive at any time, now or in the future. All surveys are anonymous.

For more information please contact the researcher by phone or email. The researchers name is Jenny Barke if you have any questions about the research, please do not hesitate to contact her via email at [jenny.barke@uwe.ac.uk](mailto:jenny.barke@uwe.ac.uk) or you can text/call on the study mobile: 07500 121137. Alternatively you can contact Jenny's supervisor, Dr Diana Harcourt on (0117) 3282192 or via email at [Diana2.Harcourt@uwe.ac.uk](mailto:Diana2.Harcourt@uwe.ac.uk).

Yours Sincerely,

CONSULTANT NAME AND DETAILS

### Appendix 28: SPSS output

(Related to page 134)

SPSS output of T-tests exploring the differences in scores on the Brief IPQ, SHS, PSQ, SCQ and BE (app) for young people by gender.

#### Group Statistics

	Gender	N	Mean	Std. Deviation	Std. Error Mean
TOTALIPQ	Female	48	48.15	11.829	1.707
	Male	20	48.65	11.385	2.546
totalhappy	Female	49	18.31	5.557	.794
	Male	19	18.26	5.248	1.204
totalpsq	Female	42	2.20	.591	.091
	Male	15	2.15	.596	.154
totalscq	Female	48	3.14	.726	.105
	Male	16	3.08	.918	.230
Total be	Female	49	19.82	10.754	1.536
	Male	18	20.61	12.329	2.906

#### Independent Samples Test

		Levene's Test for Equality of Variances		t-test for Equality of Means						
		F	Sig.	t	Df	Sig. (2-tailed)	Mean Difference	Std. Error Difference	95% Confidence Interval of the Difference	
									Lower	Upper
TOTALIPQ	Equal variances assumed	.019	.891	-.162	66	.872	-.504	3.115	-6.723	5.714
	Equal variances not assumed			-.164	36.917	.870	-.504	3.065	-6.715	5.707
totalhappy	Equal variances assumed	.426	.516	.029	66	.977	.043	1.480	-2.911	2.997

	Equal variances not assumed			.030	34.605	.976	.043	1.442	-2.886	2.972
totalpsq	Equal variances assumed	.007	.932	.246	55	.807	.044	.178	-.313	.401
	Equal variances not assumed			.245	24.525	.809	.044	.179	-.325	.413
totalscq	Equal variances assumed	.858	.358	.255	62	.799	.057	.224	-.391	.506
	Equal variances not assumed			.227	21.601	.823	.057	.252	-.467	.581
Total be	Equal variances assumed	.287	.594	-.258	65	.797	-.795	3.083	-6.953	5.363
	Equal variances not assumed			-.242	27.081	.811	-.795	3.287	-7.539	5.949

SPSS output of T-tests exploring the differences in scores on the Brief IPQ, SHS, PSQ, SCQ and BE (app) for young people by whether or not other family members were reported as having NF1.

**Group Statistics**

	Others in family with NF1	N	Mean	Std. Deviation	Std. Error Mean
TOTALIPQ	Yes	23	46.83	13.803	2.878
	No	41	49.15	10.683	1.668
totalhappy	Yes	24	18.33	4.923	1.005
	No	40	18.65	5.641	.892
totalpsq	Yes	21	2.16	.579	.126
	No	33	2.24	.595	.104
totalscq	Yes	24	3.16	.653	.133
	No	36	3.06	.861	.143
Total be	Yes	25	18.28	9.981	1.996
	No	38	21.37	12.281	1.992

Independent Samples Test

		Levene's Test for Equality of Variances		t-test for Equality of Means						
		F	Sig.	t	df	Sig. (2-tailed)	Mean Difference	Std. Error Difference	95% Confidence Interval of the Difference	
									Lower	Upper
TOTALIPQ	Equal variances assumed	1.354	.249	-.749	62	.456	-2.320	3.096	-8.509	3.869
	Equal variances not assumed			-.697	36.974	.490	-2.320	3.327	-9.061	4.420
totalhappy	Equal variances assumed	.259	.613	-.228	62	.821	-.317	1.391	-3.096	2.463
	Equal variances not assumed			-.236	53.820	.815	-.317	1.344	-3.011	2.377
totalpsq	Equal variances assumed	.204	.654	-.465	52	.644	-.076	.164	-.406	.253
	Equal variances not assumed			-.468	43.595	.642	-.076	.163	-.406	.253
totalscq	Equal variances assumed	1.589	.212	.470	58	.640	.097	.207	-.317	.511
	Equal variances not assumed			.496	56.926	.622	.097	.196	-.295	.489
Total be	Equal variances assumed	1.940	.169	-1.049	61	.298	-3.088	2.944	-8.975	2.798
	Equal variances not assumed			-1.095	58.180	.278	-3.088	2.820	-8.734	2.557

## Appendix 29: Paper Published 2013

**'It's like a bag of pick and mix - you don't know what you are going to get':**

### **Young peoples' experience of Neurofibromatosis Type 1**

#### **ABSTRACT**

**Aims** To explore the day-to-day experience of young people living with Neurofibromatosis type 1 in the UK, focusing on the role that appearance plays in this experience.

**Background** Neurofibromatosis type 1 is a genetic condition which is highly variable and unpredictable. It can result in varying degrees of visible difference (disfigurement). Both the effect of NF1 on appearance and its uncertainty can prove particularly difficult for those affected. However, very little research to date has investigated the psychosocial impact of Neurofibromatosis type 1 on young people or their experiences of managing it.

**Design** Exploratory qualitative interview study.

**Methods** Nine young people aged 14-24, with a confirmed diagnosis of Neurofibromatosis type 1, took part in semi-structured interviews between March - September 2011. Interview transcripts were thematically analysed.

**Findings** Three key themes emerged from the data: (1) 'Different things to different people' reflecting the variability of the condition; (2) 'Relationships and reactions' relating to individuals' social experience; and (3) 'Understanding and misunderstanding' reflecting participants' experiences with organisations and social structures.

**Conclusions** Findings suggest a need for further research to explore young people's adaptation and management of Neurofibromatosis type 1. In particular, raising awareness and understanding of the condition among professionals and in the general public was an important issue for young people. Additionally, access to trustworthy information about Neurofibromatosis type 1 and practical advice to support adjustment to an altered appearance and managing stigma experiences are highlighted as areas to be considered further.

#### **KEYWORDS**

Neurofibromatosis type 1, Disfigurement, Visible Difference, Appearance, Qualitative, NF1, Nursing, Adolescence, Young People,

## SUMMARY STATEMENT

### Why is this research needed?

- Neurofibromatosis type1 significantly effects psychological wellbeing and quality of life.
- The psychosocial impact of Neurofibromatosis type1 may stem from managing both the unpredictability of the condition and changes to appearance.
- Adolescence and early adulthood are periods of increased appearance salience and a time when health behaviours are consolidated, so young people with Neurofibromatosis type1 who are starting to take greater responsibility for their own health may be particularly vulnerable to the psychosocial impact of the condition.

### What are the key findings?

- Neurofibromatosis type1 can become a more prominent part of many peoples' lives during adolescence and early adulthood
- Particular challenges that young people with Neurofibromatosis type1 describe include managing other people's reactions to their appearance, living with the uncertainty of the condition and the general lack of awareness about it.

### How should the findings be used to influence practice and/or policy?

- Evidence based interventions, including social skills training, are needed to support young people affected by appearance-related concerns associated with Neurofibromatosis type1.
- Young people with Neurofibromatosis type 1 need easily accessible, reliable information and social support to help them manage the demands of the condition.
- Raising awareness and understanding of Neurofibromatosis type1 amongst health professionals could usefully support young peoples' overall experience of the condition.

## INTRODUCTION

Neurofibromatosis type 1 (NF1) is a highly variable and unpredictable genetic condition with an incidence rate of 1: 2500 - 1:3000 (Ferner et al 2007). Diagnosis is based on clinical assessment and visible signs of the condition include café au lait spots (coffee coloured birthmarks), neurofibromas (benign tumors on the skin) and plexiform neurofibromas (diffuse tumors that grow along a nerve). Other features of NF1 can include Lisch nodules of the iris (small pigmentation in the iris which causes no disturbance to vision), axillary freckling (freckling/pigmentation in groin and armpits), optic

gliomas (tumor of the optic nerve) and skeletal complications including scoliosis (curvature of the spine) and pseudoarthrosis (meaning false joint). Macrocephaly (larger head size) is also common and short stature is found in around a third of people with NF1 (Hersh 2008).

Cognitive deficits are a common complication in children with NF1; between 30-65% of children with the condition may have learning difficulties (Cutting et al, 2004) and behavioural problems. Learning difficulties, Autistic Spectrum Disorders (ASD) and Attention Deficit Hyperactivity Disorder (ADHD) are more common in people with NF1 than in the general population (Barton and North 2004, Ferner et al 2007). Up to 50% of people with NF1 meet the criteria for diagnosis of ADHD (Gilboa et al 2011) and this may undermine both their academic achievement (Pride et al 2012) and social functioning (Barton and North 2004), and this impact has been found to persist into adulthood (Mautner et al 2012).

Research demonstrates that NF1 can have a significant effect on quality of life and psychological adjustment (Mouridsen & Sorensen 1995; Zoller & Rembeck 1999; Samuelsson & Riccardi, 1989; Graf et al 2006; Krab et al 2009; Noll et al 2007). Adolescence and early adulthood may be a particularly challenging time for people with NF1, since this is often a time during which appearance concerns are heightened (Rumsey & Harcourt, 2012), yet this is when neurofibromas typically start to develop. Furthermore, young people with NF1 have been identified as having poor social skills (Barton & North, 2004), difficulties processing social information (Huijbregts et al 2010) and forming friendships (Noll et al 2007). While this may be explained in part due to the high prevalence of ADHD, children with NF1 have been found to have problems with attention and social information processing even without the presence of ADHD (Huijbregts et al 2010). Poor social skills could be considered a risk factor in terms of a young person's ability to manage the appearance-related challenges presented by NF1 because social skills are important in mediating the effects of an altered appearance (Rumsey & Harcourt, 2012). The unpredictability of the appearance changes associated with NF1, and a body of literature that has shown that the extent or severity of a visible difference is not associated with levels of distress, (Moss, 2005, Ong et al 2007) suggest that appearance concerns are likely to be as much an issue for people with no, or few, physical signs of NF1 as they are for those with more noticeable changes.

Commonly reported challenges for people living with a visible difference of any kind include difficulties forming relationships, negative reactions from others and discrimination (Thompson & Kent 2001, Kent 2005). Yet, despite the potential psychological challenges for young people with NF1, there may also be positive consequences; family relationships may be strong and provide valuable support that is a key factor in a young person's psychosocial adjustment (Graf et al 2006, Krab et al 2009).

## **Background**

Despite the potential impact (both positive and negative) of NF1 on psychosocial adjustment during adolescence and early adulthood, limited research has examined the lived experience of this age group. Instead, much of the available research relies on adults' retrospective reports or parental reports of their child's experiences, yet parents' and young people's concerns and reports differ, with

parents more pessimistic on measures such as social inclusion (Sebold et al 2004, Wolkenstein et al 2008). Some research has involved younger children, who may be without obvious signs of NF1 since changes to appearance are often not apparent until after puberty (Barton & North, 2007). Furthermore, much of the limited research reported as being with young people or adolescents includes a young sample. For example, both Graf et al (2006) and Counterman (1995) report the mean age of participants in their studies of adolescence as being 11 years, whilst Huijbregts et al (2010) report a mean age of 12 years. Sebold et al (2004) report a mean participant age of 15 years and, interestingly, they report differences between those under and over 15 years of age suggesting that young people's perceptions of their condition may change with age. It is also important to note that findings from studies with young adults (Hummelvoll and Antonsen, 2013) have reported differences between the youngest participants aged 18-25 and those aged 26-37 in terms of friendships, depressive difficulties and self-confidence. In summary, the limited available research suggests adolescence and early adulthood are times during which young peoples' perceptions of their condition change.

As perceptions alter, it is important that young people take greater responsibility for their own healthcare and transition positively from paediatric services to adult care. Yet a recent study with young adults with NF1 (Oates et al, 2013) reported that most participants had not had a NF1 health check since transition to adult healthcare, few had a good understanding of NF1 and many had not sought medical advice as difficulties arose. These are factors which Oates et al (2013) suggest can lead to increased NF1-related complications. Given the variability and unpredictability of NF1, there is no single best approach to caring for people with the condition, but the main aims of care should be to support adjustment and provide the best possible treatment for specific complications (Rubenstein and Korf, 1990).

In light of the gap in the existing literature, the current study specifically investigated young peoples' experiences, concentrating on individuals aged between 14 and 24 and exploring both the challenges and any positive experiences of living with NF1.

## **THE STUDY**

### **Aim**

This study aimed to obtain an in-depth understanding of young peoples' day-to-day experience of living with Neurofibromatosis type 1 focusing on the role that appearance plays within this experience in order to inform the provision of support for young people with the condition.

### **Design**

The design reflects the researcher's exploratory approach towards this under-researched area and the desire to ensure young people were empowered and heard from directly. Furthermore this approach recognises that NF1 is a highly variable condition and as such young people may have many diverse symptoms and experiences. A qualitative approach using visual methods was therefore

chosen as a way of gaining rich in-depth accounts of young people's different and complex experiences (Rich & Ginsburg 1999).

### Participants

Nine young people with NF1 were recruited through the Neuro Foundation newsletter (a UK-based support group for people with Neurofibromatosis), the online forum of Changing Faces (a charity offering support for people living with disfigurement), the Centre for Appearance Research website and advertisements posted on social media sites related to NF1. Participants ranged in age from 14 (the age at which neurofibromas commonly appear) to 24 years (in line with the World Health Organisation's upper definition of youth) in order to obtain a current account of experiences during this life stage. See table 1 for participant details.

<b>Participant pseudonym</b>	<b>Gender</b>	<b>Age</b>	<b>NF1; Inherited or new mutation</b>	<b>Highest educational level to data</b>
Tina	Female	21	Unsure	University
Sarah	Female	23	New	Postgraduate
Joanna	Female	17	New	College
Robert	Male	20	New	College
Ros	Female	24	Inherited	University
Daniel	Male	24	Inherited	Postgraduate
Katie	Female	14	Inherited	School
Lucy	Female	16	Inherited	School
Mark	Male	21	New	University

Table 1: Participant details

### **Data collection**

Young people who were interested in taking part contacted the first author and were sent an information pack (including parental information if they were aged under 18). Those who wanted to participate were invited to contact the researcher to arrange an interview. Innovative and visual methods have previously been used in appearance research as a way of engaging people in a potentially sensitive topic (Rumsey & Harcourt 2012; Coad 2007), so participants in this study were offered the opportunity to bring photographs to their interviews or to use timelines during the interview. Using a timeline entailed the participant drawing a line on a piece of paper with their date of birth at one end and the current date at the other, and then adding significant dates and events in relation to having NF1 between the two. The photographs and timelines themselves were not analysed but were used as an empowering way for young people to share their experience (Sebold et al 2004; Metcalfe et al 2011).

An interview guide was developed through a review of literature around engaging young people in research, NF1 and appearance research, and with input from people working with young people with NF1. Topics included the impact of NF1 on the young person's life, the role of friends and family, knowledge of NF1 and experiences of treatment and support. An assessment of appearance was not recorded as previous research has suggested it is subjective rather than objective assessment of appearance that predicts wellbeing (Moss 2005, Ong et al 2007), and participants were specifically asked about their subjective experience of their appearance during interviews. Eight interviews were conducted face-to-face and one by telephone (Ros), at the participant's choosing. Interviews were conducted by the first author and face-to-face interviews took place in participants' homes (n= 6), or another setting of their choice (n= 2). Interviews lasted between 30 and 80 minutes and were digitally audio recorded.

### **Ethical considerations**

Ethics committee approval was obtained from the faculty research ethics committee at the first author's host institution. Informed consent was obtained from all participants and parental consent for participants under 18. Participants were reminded that interviews could be stopped or paused at any time and that they did not have to answer any questions if they were uncomfortable doing so, or were uncertain of their answer.

The researchers were conscious that some participants may not have in-depth knowledge of NF1, and were therefore careful to ensure the interview process did not introduce areas of concern to young people for the first time. Participants were therefore not asked directly about issues such as future prognosis or heritability/reproductive decision making until the subject was mentioned by the participant themselves. While this may have meant some participants were not asked some questions on the interview guide, it was felt this was the most ethical way to approach these circumstances.

## Data analysis

Interviews were transcribed verbatim and field notes were kept by the researcher after each interview. These were not analysed but were used as a reflexive tool to support the analysis of interview transcripts. Since NF1 is a highly variable condition and participants could be expected to have a multitude of different experiences, thematic analysis was chosen because it maintains the richness of data whilst allowing areas of commonality to be analysed. All transcripts were coded line-by-line and codes were developed into subthemes and themes, following the recommendations of Braun and Clarke (2006).

## Rigor

To ensure rigor of analysis, interviews were transcribed verbatim in a timely manner and individual transcription codes, sub themes and themes were verified throughout the research process, all themes and interpretations made by the first author were reviewed by the second and third authors and were discussed until there was a consensus (Morse et al 2002).

## Findings

The findings presented are based upon interviews with nine participants (6 female; 3 male) who all lived in the United Kingdom and ranged in age from 14 to 24 years. Five participants chose to use timelines or brought photographs to their interviews, which were used as outlined above.

Central to all their accounts of their experience of NF1 during adolescence and early adulthood were the variability, unpredictability and visibility of the condition. Its effect on appearance was discussed in terms of adapting to a changing appearance and managing interactions with other people. Analysis revealed three key themes: (1) 'Different things to different people' (2) 'Relationships and reactions' and (3) 'Understanding and misunderstanding'

### **(1) Different things to different people**

NF1 can be characterised by its variability (Ablon 1999, Ferner et al 2007). This was particularly evident in how young people described the condition. Some described it as a skin condition, some talked of 'tumors' and 'lumps', some referred to how it affected a specific part of their body (eg legs), and others discussed learning difficulties or ADHD. Participants who chose to use visual methods detailed a range of symptoms and treatments on timelines and brought photos with them that depicted diverse symptoms and their hospital stays. As Daniel commented: *"It's like a bag of pick 'n' mix {a reference to a common practice in UK sweet shops where people buy a few of several different types of sweets} you don't know what you're going to get really, it could be anything"*

Accounts also varied regarding the emotional effect of NF1. Some participants felt it held little significance and that there was no real emotional effect at all, whilst others felt it had an extremely profound influence on their life. For instance, Robert commented that NF1 *"Bothers me now and again but it doesn't put me too down"* whereas Ros explained that *".....if I didn't have NF I wouldn't have the problems I have got"*.

Participants talked of adolescence being a time of change, a time of re-evaluating or learning about NF1 as they came to understand what it meant for them, and the influence it might have on their lives throughout adulthood. Whilst they had varied experiences and symptoms, many felt their understanding and feelings about NF1 had changed during adolescence. Katie, the youngest participant, described a growing awareness of having NF1 in recent years *“I don’t know if I knew when I was younger or not and I have certainly become more self conscious of it”*. Older participants reflected that their own increased awareness had led them to want to know more about the condition. For some, the desire to learn more stemmed from physical changes:

*“I think it was sort of early years of secondary school when I started to notice a few more of lumps and just a bit more curious about it and see how it was affecting me, and I yeah so I got a few more like information packs on it and stuff” (Mark).*

Most participants talked of how their conceptualisation of the condition altered as they learned more about it. For example, Sarah was treated primarily for pseudoarthrosis during her childhood, and it was only as she got older that she realised this was part of a genetic condition and learnt more about it:

*“I think when I was younger it was just the leg stuff that used to bother me, now it is more like the neurofibromas and stuff..... it had always been a leg thing and then to kind of reconceptualise that, it was quite weird .....it was just I’ve got NF that means my legs and my shin bones don’t work properly so ....there’s been a lot of learning since then” (Sarah).*

## **(2) Relationships and reactions**

Participants felt that relationships were affected by NF1 in a myriad of ways. This was discussed with reference to (a) *Family*, (b) *Friends* and (c) *Other peoples’ reactions*

### *Family*

NF1 was described as a ‘family thing’; a diagnosis affected the whole family. Most participants discussed how their families seemed to understand the condition, and were their main source of support and information. Family members who had NF1 themselves were thought to understand the condition and its impact more than those without it.

*“... I think myself and my dad can understand each other more, where you know, people without NF can’t really” (Ros)*

### *Friends*

Consistent with previous research with young people with an altered appearance (Williamson et al, 2010); friends were an important source of support, understanding and trust. Some participants found telling friends about NF1 straightforward:

*“it just takes two minutes of conversation and then you don’t need to talk about it again” (Mark)*

However, others discussed how they avoided talking about NF1 with others:

*“I wouldn’t want to say NF because my fear is they’ll go onto Google or Wikipedia and then find this kid has varying disabilities this kid may have a fit and I don’t want them to think that” (Daniel)*

*Other peoples’ reactions*

Participants discussed managing situations where strangers had stared at them or asked questions about their appearance. Some found this distressing, whilst others had learnt to manage it:

*“I am always scared that people are looking at you, are they staring at you?” (Tina)*

*“I’ve never really been too uncomfortable because I’ve grown up with it, so from an early age I’ve always been used to having to say something.” (Mark)*

Most participants reflected that their concerns about other people’s reactions to them could make social situations difficult, and in some circumstances cause them to avoid interacting with others:

*“...when I meet new people, there is immediately this thing where, oh she walks a bit different, her legs are a bit weird, she’s just tripped over her own feet, like you know, and it’s like I just feel immediately awkward about all of those things and I think.....just screw it I won’t even bother trying to talk to anybody.” (Sarah)*

### **(3) Understanding and misunderstanding**

This theme relates to participants’ experiences with organisations including healthcare, school, support groups, and the media, and the influence these organisations had on relationships and psychosocial adjustment.

Whilst it was very important to participants that health professionals were available to give good quality information and advice about NF1, most commented that it was poorly understood within the medical community. For example, when Tina drew her timeline she added several occasions when health professionals had not understood her condition. She highlighted a specific occasion when she had asked her GP for information and was frustrated that he had to look up the condition, *“they looked back on my file and said ‘hang on we have got neurofibromatosis here’.....the doctor was reading like a massive big book in front of me and I was like ‘what are you doing mate, come on’”*

Those who described positive relationships with health professionals commented on their perceived expertise and professionalism. In particular, those who had seen specialist nurses, doctors or Neuro Foundation specialist advisors described being very happy with the advice and care they had received. Experts played a crucial role in putting information into context and some of the participants added contact with experts to their timelines.

A few participants felt NF1 had positively affected their education. Katie explained *“with NF I get to go to college and I’ve got experience before people at school”* referring to the opportunities she had had to go on specialist work experience programs at a younger age than many of her peers. Others talked of a ‘battle’ to get the support they needed and to have their NF1 and any associated learning difficulties recognised, and felt this had negatively affected their education. Medical professionals could be particularly important in supporting young people at school, as Ros explains:

*“I struggled all the way through primary school up until secondary school. It was only until secondary school where my paediatrician and he stepped in and said this girl needs you know help in her work and he arranged for an educational psychologist and that’s when I got found out that I obviously had dyslexia” (Ros).*

The Neuro Foundation was an important resource for some of the young people in this study, four of whom had been to residential camps or events run by the organisation. They described these as being supportive, particularly because they were designed for their age group and gave them an opportunity to meet others of a similar age and experience to themselves.

Some participants preferred to use online support rather than face-to-face groups, and as shown in the following quote, were sometimes pleased to speak to others without the need to focus on their condition:

*“I used the instant messaging thing which was really good because there would be loads of people there and you’d be talking to lots of different people and I’d kind of just go on and wouldn’t always really talk about NF it would just be a shared understanding” (Sarah)*

The way in which NF1 is represented in the media (such as the internet, television and in print) was important to the participants. Most thought it is misunderstood or unknown to the general population, and they were worried and frustrated by the quality of information available, and the negative and pathologising way NF1 is portrayed.

*“...if I see any articles regarding NF, 9 times out of 10 they got it wrong. That really winds me up.” (Daniel).*

Sarah described how inaccurate reporting of NF1 impacts on her experience of trying to minimise the impact of the condition on her life;

*“.....what if my friends have read that and thought oh that’s what {Sarah} has got as well, that Elephant Man disease or whatever. That was, that was really hard because I’m trying to.... I was trying to cope with the things that made me different and trying to make myself as normal as possible and deal with all these things that made me different and then to have those horrible stories like thrown in the mix.” (Sarah)*

## DISCUSSION

This study explored young peoples' experiences of NF1 and has given an insight into their views about the condition and its effect on their lives. Findings support previous suggestions (Ablon 1999, Ferner et al 2007) that NF1 may be particularly challenging due to its unpredictable nature and its effect on appearance. Young people face these challenges within a society that holds a robust belief about the importance of appearance and the stereotypical view that "what is beautiful is good" (Dion et al, 1972) and during a life stage when body image concerns are likely to be emphasized (Levine & Smolak 2004). Additionally, findings suggest the lack (or perceived lack) of awareness and understanding of NF1 may make adjustment all the more complicated.

Whilst participants described the pressures and difficulties they faced as a result of having NF1 (particularly other people's reactions to any visible signs of the condition, their concern over the uncertainty of dermatological changes and the general lack of awareness of the condition), it is important not to pathologise the experiences of people living with an unusual or altered appearance (Rumsey & Harcourt 2004, Egan et al 2011) and to highlight that the participants in this study displayed considerable resilience and were living positively with NF1 despite facing these numerous challenges. For example, as suggested in previous research (Ablon 1999, Ferner et al 2007) adolescence was, for many, a time during which the condition became a more prominent part of their life, partly due to physical changes (condition specific or pubertal), but also as they reconceptualised their understanding of the condition. This reconceptualisation, alongside their desire for more information as their symptoms change, suggests adolescence may be a time during which availability of good quality information and access to specialists is crucial to support their adjustment to the condition. Yet, as young people make the transition into adulthood and start to use adult healthcare services it may be that they receive less specialist care and have less access to specialist information and support (Oates et al, 2013). Health professionals need to recognise the possibility of appearance concerns amongst this patient group, thus legitimizing young person's concerns (Clarke 1999). Since the development of self-esteem and the formation of lifelong supportive coping mechanisms are formed during adolescence (Holmbeck 2002), this is a time of opportunity to support the development of positive coping and resilience (Ahern et al 2008).

The young people in this study called for a greater awareness of NF1 amongst health professionals, education specialists and the general public, and, as previous authors (Dheensa & Williams 2009) have suggested they felt that mistruths such as the media's persistent tendency to link NF1 with 'the elephant man' should be challenged (Ablon, 1999).

In line with the findings of studies with young people with other appearance-altering conditions (Thompson & Kent 2001, Thompson & Broom 2009, Williamson et al 2010), participants valued social support from friends, family and support groups, and practical information to help them manage other peoples' reactions to their appearance. Adolescence is a transitional point with regards to young peoples' social skills (Pitt, 2009), and managing other people's questions and reactions and explaining NF1 to them was an important concern for the young people in this study. Social skills training has

been shown to be beneficial for many people with a visible difference (Rumsey & Harcourt, 2007, Clarke 1999) and, given that young people with NF1 may have poorer social skills than their non-affected peers (Barton & North, 2007), this would seem to be a potentially very supportive and beneficial intervention for this group.

Young people can find it difficult to explain NF1 to others (Hummelvoll & Antonson 2013; Sebold et al 2004), partly because of the variability of the condition. In addition, some young people in the current study were reluctant to discuss NF1 because of their concerns that others would search for the condition on the internet, see the worst case scenarios and then classify them in this way. Helping young people find ways to talk about NF1 in a simple and positive manner may be particularly useful during adolescence and early adulthood.

### **Limitations**

There are some limitations to this study. Considering the variability of NF1, including the possibility of associated behavioural and learning difficulties, it is not possible to generalise the findings from this study to other young people with the condition. However, while the sample size is small, they are appropriate for the qualitative methods we have employed and have provided a rich and in depth account of young people's experience of NF1.

### **CONCLUSION**

In conclusion, this study has offered an original insight into young people's experiences of living with Neurofibromatosis type1, and has identified four areas warranting further examination and which may provide benefits for those affected. First, a need for increased awareness and understanding about NF1 amongst the general public and professionals such as teachers and health professionals. . Second, ensuring young people have easy access to trusted information about NF1, including professionals with appropriate expertise. Third, easy access to social support that is age-appropriate (this could be in person and/or through online resources). Finally, a range of psychosocial and psycho-educational interventions to support the development of social skills and adjustment to an altered appearance.

Until now very little research has specifically examined the experiences of young people living with NF1. Whilst this study starts to explore this overlooked area, more research is needed in order to support clinicians looking to provide the best possible care for people with NF1 during adolescence and into early adulthood in order to positively support the development of resilience.

### Appendix 30: List of presentations undertaken to disseminate findings

Barke, J., Harcourt, D., Coad, J., (2013) "It's like a bag of pick and mix – you don't know what you're going to get": Young peoples' experiences of neurofibromatosis type 1, surveys of parents and young people, Division of Health Psychology Conference, Brighton, September 2013 (Poster)

Barke, J., Harcourt, D., Coad, J., (2013) Young people's experience of neurofibromatosis type 1: A survey of young people, parents and health professionals, Childrens Tumor Foundation Conference, USA, June 2013 (Poster)

Barke, J., Harcourt, D., Coad, J., (2013) Young peoples' experience of NF1; The parents' perspective, NF Knowledge Exchange Day, Manchester Metropolitan University, Manchester (Poster)

Barke, J., Harcourt, D., Coad, J., (2012) Young peoples' experience of NF1; Interviews with young people, 15th European NF meeting, Istanbul (Oral)

Barke, J., Harcourt, D., Coad, J., (2012) Young peoples' experience of NF1, Appearance Matters conference, hosted by Centre for Appearance Research, Bristol (Oral)

Barke, J., Harcourt, D., Coad, J., (2012) What do young people think about NF1? Neuro Foundation AGM, Birmingham (Oral)

Barke, J., Harcourt, D., Coad, J., (2011) Young peoples' experience of neurofibromatosis type 1: interviews with health professionals, Division of Health Psychology Conference, Southampton (Poster)

Barke, J., Harcourt, D., Coad, J., (2011) Young peoples' experience of neurofibromatosis type 1: The health professionals' perspective, British Society for Human Genetics Conference, Warwick (Runner-up prize for poster presentation)

Barke, J., Harcourt, D., Coad, J., (2011) Young peoples' experience of NF1: why appearance is important, PsyPAG conference (Psychology post graduate Group) Bangor (Runner-up prize for oral presentation)

Barke, J., Harcourt, D., Coad, J., (2011) Young peoples' experience of NF1: an exploration of psychosocial and ethical issues, medical genetics forum at the Institute of Medical Genetics at University Hospital of Wales, Cardiff (Oral)

Plus presentations at UWE Psychology department and postgraduate events (2010, 2011, 2012 and 2013), a Great Ormond Street Hospital Genetics Team Meeting (2012) Great Ormond Street Hospital, London, and the Neuro Foundation Medical Board, Guys Hospital, London (2010).