Falling short: The psychosocial impact of living with Russell-Silver syndrome.

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A thesis submitted in partial fulfillment of the requirements of the University of the West of England, Bristol for the degree of Professional Doctorate in Health Psychology

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“For a few seconds Oskar saw through Eli's eyes. And what he saw was... himself. Only much better, more handsome, stronger than what he thought of himself. Seen with love.”

- John Ajvide Lindqvist (2005) Let the Right One In
Thesis introduction and systematic review

This thesis forms part of the research competency, one of five competencies, required for the fulfillment of a professional doctorate in health psychology. The research competency had two elements, the systematic review and an empirical study including a reflective chapter. The introductory chapter of the empirical study is usually based upon the systematic review.

The empirical study explored the lived experience of the rare genetic condition Russell-Silver syndrome. Through in-depth, semi-structured interviews, and thematic analysis, what it is like to live with this syndrome was elucidated. The systematic review was titled ‘Increasing the uptake of stop smoking support in secondary care: A systematic review’ and through narrative synthesis aimed to determine the most effective method of increasing the uptake of stop smoking support by smokers in secondary care.

When planning the systematic review, I intended to base my empirical study on the findings. Working as a smoking cessation advisor for Solent NHS Trust, I was ideally placed to conduct this research. But after some consideration I decided to broaden my experience beyond smoking cessation and looked for a different research opportunity. I was fortunate enough to find and be offered a new position in University Hospital Southampton NHS Foundation Trust with a team researching Russell-Silver syndrome. As my systematic review does not inform my empirical study I have included it in appendix 16 and have reviewed the literature for Russell-Silver syndrome and related areas in the introduction chapter of this thesis.
# Table of contents

Abstract .............................................................................................................................................. 7

Acknowledgements ........................................................................................................................... 8

1.0 Introduction ................................................................................................................................... 9

1.1 Russell-Silver syndrome .................................................................................................................. 9
  Table 1. Clinical features of Russell-Silver syndrome .................................................................... 10

1.2 Treatment ................................................................................................................................... 10

1.3 Living with short stature ................................................................................................................. 11

1.4 Achondroplasia, Turner syndrome and Constitutional growth delay ........................................ 13

1.5 Visible Difference and appearance-related concerns ............................................................... 17

1.6 Common Sense Model of self-regulation of health and illness
  Figure 1. The five domains of illness representations (Leventhal et al., 2003). ............................ 77

1.7 Conclusion ..................................................................................................................................... 21

1.8 Aim .............................................................................................................................................. 22

1.9 Research questions ........................................................................................................................ 22

2.0 Methodology .................................................................................................................................. 23

Table 2: Ontologies and epistemologies in psychology (adapted from Braun and Clarke (2013)) ......................................................................................................................... 24

2.1 Qualitative methodology .............................................................................................................. 25

2.2 Data generation ............................................................................................................................. 26

2.3 Method of analysis ........................................................................................................................ 26
  Figure 2. Key stages of Braun and Clarke’s TA analysis (2006, 2013) ........................................ 28

2.4 Researcher influence on data generation and analysis ............................................................... 29

3.0 Method ......................................................................................................................................... 31

3.1 Participants .................................................................................................................................. 31
  Table 3. Demographic information .............................................................................................. 31

3.2 Recruitment strategy .................................................................................................................... 32
  Figure 3. Diagram of recruitment .............................................................................................. 33
3.3 Interview schedule ........................................................................................................... 33
3.4 Procedure .......................................................................................................................... 34
  3.4.1 Data generation ............................................................................................................. 34
  3.4.2 Consent ......................................................................................................................... 35
  3.4.3 Transcription/management of data ............................................................................ 35
3.5 Data analysis ..................................................................................................................... 36
3.6 Ethical issues ..................................................................................................................... 37
  3.6.1 Confidentiality .............................................................................................................. 37
  3.6.2 Ethical dilemmas .......................................................................................................... 37
  3.6.3 Debriefing .................................................................................................................... 35
  3.6.4 Researcher safety ......................................................................................................... 38
  3.6.5 General ethics ............................................................................................................. 38
3.7 Quality in qualitative research ......................................................................................... 38
  Table 4. Eight “Big-Tent” Criteria for Excellent Qualitative Research (Tracy, 2010) ............ 40
4.0 Findings ............................................................................................................................. 42
  Table 5. Participant age range, height and treatment. ............................................................ 42
  Figure 4. What is it like to live with RSS? A conceptual map of themes and sub-themes. .... 43
4.1 “It’s not just all about height” ........................................................................................... 43
  Table 6. Characteristics of RSS that had the biggest impact on participants and unprompted appearance-related concerns......................................................... 44
Partici
  4.1.1 Summary .................................................................................................................... 49
4.2 Everyone’s comparing, everyone’s judging ................................................................. 50
  4.2.1 Summary .................................................................................................................... 62
4.3 ‘Mayor of the friend zone’ ............................................................................................... 62
  4.3.1 Summary .................................................................................................................... 70
5.0 Discussion .......................................................................................................................... 71
5.1 Key discussion points from findings ............................................................................ 72
  5.1.1 Adolescence was a difficult time ............................................................................... 72
  5.1.2 “You don’t have Russell-Silver anymore; it’s only a childhood condition” .............. 75
  5.1.3 A mismatch between healthcare provision and patient need .................................. 79
  5.1.4 What patients, families and health professional need to know about living with RSS... 81
Abstract

We do not know what it means to live with Russell-Silver syndrome (RSS), as currently there is a dearth of literature exploring the lived experience of this rare genetic condition characterised by pre and postnatal growth retardation. Discovering how this syndrome affects the lived experience will provide valuable information for better healthcare provision and aid families in making difficult treatment decisions. In this qualitative study, in-depth semi-structured interviews were used with 15 participants (6 female). Using thematic analysis, three themes were identified: “It’s not just all about height”, ‘Resilience - threat and preservation’, and “mayor of the friend zone”. These themes describe participants’ struggles with varied psychosocial, appearance and body image related concerns. The key findings were: participants experienced appearance-related concerns that surpassed a concern about height; adolescence was a particularly difficult time; and there was a mismatch between patient need and existing healthcare provision. Two main recommendations were made: Firstly, psychosocial evaluation and interventions to improve self-esteem, self-confidence, negative body image, and social interactions during early adolescence could ameliorate psychological distress. And secondly, a conflation of the 2016 expert consensus statement, which summarised recommendations for the management of patients with RSS as well as for diagnosis and investigation, and the results from the present study is needed to inform a care pathway recommending psychosocial assessment and that continues into adulthood.
Acknowledgements

First and foremost, I would like to say a huge thank you to all the participants who shared their experiences with me. Secondly, I would to thank my supervisors - Angela and Liz - who formed the best supervisory team I could have hoped for. You have encouraged me, filled me with confidence, and, without you this would have been a much harder journey. Thirdly, I would like to thank Aaron who has given me a much-needed distraction from my studies and reminds me that there is more to life than work. Fourthly, I would like to thank my family: Mum, Dad, Pesh, John, and Donna. You have always supported and encouraged me in all my endeavours and have been there no matter what. Fifthly, I want to thank my ‘study-buddy’ Nisha, without whom this would have been a lonely journey. And finally, I want to say thank you to the 2013 professional doctorate in health psychology cohort; it was a pleasure studying with you and I wish you all the best!
1.0 Introduction

This qualitative study was nested in a larger study, which aimed to ascertain the long-term health outcomes for people with Russell-Silver syndrome and to establish whether growth hormone therapy was effective in increasing height. The larger study recruited 34 participants, and the participants in my study were recruited from this group.¹

1.1 Russell-Silver syndrome

Russell-Silver syndrome (RSS), also known as Silver-Russell syndrome, is a rare genetic condition caused by chromosomal problems (Binder et al., 2011). It is characterised by slow growth in the womb leading to poor post-natal growth, short stature in adulthood, triangular facial appearance, asymmetry, feeding difficulties (Wakeling, 2011), low muscle mass and poor muscle function (Schweizer et al., 2008). The prevalence of RSS is not easily determined as making a clinical diagnosis is difficult due to ill-defined features that vary in severity (Christoforidis et al., 2005, Wakeling, 2011). However, the published incidence of RSS ranges from 1:3,000 to 1:100,000 around the world (Price et al., 1999), with males and females equally affected (Binder et al., 2011). Currently there are two known molecular abnormalities that cause RSS: a loss of DNA methylation at the H19 locus causes 60% of cases (this epigenetic mutation causes a reduction in a growth-promoting factor called IGF2); while maternal uniparental disomy of chromosome 7 (MatUPD7, the inheritance of both chromosome 7s from the mother with no contribution from the father) causes 5%-10% of cases (Netchine et al., 2007). The remaining patients have clinical features but an unknown molecular cause (Wakeling, 2011). Table 1 summarises the clinical characteristics of RSS and the prevalence of each characteristic.

¹ I made the decision to write in the first person. Writing in the first person engages the reader and brings them closer to the author and the voice of the participants (Gilgun, 2005). This also serves to instil my ownership of the research, which is of particular importance as it was part of a wider study.
Table 1. Clinical features of Russell-Silver syndrome.

<table>
<thead>
<tr>
<th>Clinical Feature</th>
<th>Description</th>
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<tbody>
<tr>
<td>Short Stature</td>
<td>Children with RSS often show normal growth throughout childhood, but typically no catch-up growth, resulting in a final height -4.2 SD below the mean in adulthood and an average height of 140cm (4ft 7in) in women and 151cm (4ft 11in) in men (Binder et al., 2011).</td>
</tr>
<tr>
<td>Asymmetry</td>
<td>Asymmetry can affect the body, face and/or limbs. Prevalence varies from about one third upwards (Abraham et al., 2004, Price et al., 1999, Wakeling, 2011, Wakeling et al., 2010). Wakeling et al. (2010) found facial asymmetry in 37% of patients.</td>
</tr>
<tr>
<td>Learning Disabilities</td>
<td>In a sample of 50 children and adults Price et al. (1999) found that, those of school age and above, 37% (14) had received a statement of special educational needs, which was similar to the prevalence found by Lai et al. (1994). Patients with MatUPD7 are more likely to have mild learning disabilities than patients with H19 (Wakeling, 2011).</td>
</tr>
<tr>
<td>Delayed Development</td>
<td>Wakeling et al. (2010) described global developmental delay (gross and fine motor skills, speech and language, social and emotional skills and cognitive skills) in 34% of their sample, but said that severe delay was uncommon.</td>
</tr>
<tr>
<td>Feeding Problems</td>
<td>Feeding problems in children with RSS generally stop around the age of six (Blissett et al., 2001) and affect 77% of children (Christoforidis et al., 2005). There is no documented account describing the impact of these early problems affecting adults.</td>
</tr>
<tr>
<td>Cleft palate</td>
<td>This is described as a common characteristic (Saal, 1993).</td>
</tr>
</tbody>
</table>

1.2 Treatment

Families often have to make the difficult decision to start their young child on growth hormone therapy which is used to improve final height of children with growth failure (Bryant et al., 2007, Wakeling, 2011). Although this treatment does not appear to have any notable physical side effects (The MAGIC Foundation, 2016), it does involve an injection under the skin six to seven times a week for several years, until final height is reached (Bryant et al., 2007). Although growth hormone therapy is the main treatment for RSS, treatment options for other characteristics of RSS include speech therapy for oral motor dysfunction (Price et al., 1999), operations to correct finger curving (Lahiri and Lester, 2009) or overcrowded teeth (Price et al., 1999), limb-lengthening surgery to correct asymmetry (Abraham et al., 2004), dieticians (Wakeling, 2011), and feeding tubes into the nose or stomach for feeding difficulties (Christoforidis et al., 2005). Families of children with RSS would be able to make more informed decisions about treatment if more was known about how this syndrome affects the lived experience throughout a person’s life.
Clinical features help build a picture of how RSS affects the individual. However, there is only a small amount of literature investigating what it means to live with the condition, providing little detail about how the characteristics of RSS impact the patient or their family. I found two examples: Wakeling (2011) observed that those with limb asymmetry in her sample all had scoliosis (abnormal curvature of the spine) and suffered pain as a result; and Abraham et al. (2004) described the implications of asymmetry for their oldest patient who used a wheelchair and walker to remain mobile. The literature also lacks insight into the lived experience of RSS in adulthood. The most recent expert consensus statement for RSS, which summarised recommendations for the management of the condition as well as for diagnosis and investigation, stated that though only a small number of adults are followed up, those that are report “few medical problems” (Wakeling et al., 2016, pp. 16); there was little consideration of the psychosocial impact of the condition. In contrast to these limited examples in the RSS literature, the research on short stature in general gives an indication of the challenges of living with restricted growth.

1.3 Living with short stature
Research suggests that people with short stature can often be dissatisfied with their appearance. Cooke (2004) found that men and women aged between 19 and 22 with short stature, who were born prematurely, rated themselves as less attractive than a control sample of people who were not born at full term but who were within average height ranges. Short stature can predict negative body image and is often found to be the case in boys more so than girls; problems can begin when an individual’s body image deviates from the cultural ideal. For example, being a taller man rather than shorter is often seen as more desirable and taller men may receive more favourable attention (Vilhjálmssson et al., 2012). But does dissatisfaction with height have a psychosocial impact or affect quality of life?

Voss published data from the Wessex growth study, along with many articles reviewing the literature regarding short stature, concluding that short stature has negligible effects on quality of life and psychosocial factors, and inferred that short stature, as a stand-alone characteristic, does not appear to have any discernible disadvantages (Sandberg...
and Voss, 2002, Voss, 1995, Voss, 1999, Voss and Sandberg, 2004, Voss, 2001, Voss, 2006). The Wessex growth study, a longitudinal study comparing short ‘normal’ children to children of average stature, measured children’s self-esteem and body image over a period of eight years and found no significant difference between groups at aged eleven to thirteen (Downie et al., 1997). Voss’s participants were all “short but otherwise normal” children (Voss, 1999, pp. 370), meaning a child that had no medical reason for their short stature. These findings were replicated in the same sample once participants reached young adulthood (18-20) (Ulph et al., 2004). Voss and colleagues identified that even though adolescents with short stature are dissatisfied with their height and would choose to be taller, this does not result in psychological distress or any disadvantage (Voss, 1995). Naiki et al. (2013) made the same conclusion from a sample of children with idiopathic short stature.

Voss (1999) and (Naiki et al., 2013) criticised studies that found short stature led to disadvantage and psychosocial issues on the basis that participants were drawn from clinical populations where those with the greatest difficulties and parental concerns would be found. This indicates that there may be a noteworthy difference between people who are short in the absence of any other health issues and those with, for example, idiopathic short stature. Children with idiopathic short stature undergo many medical investigations to ascertain the aetiology of their stature, but no genetic cause or disease is identified (Bryant et al., 2007). Children with idiopathic short stature are often treated with growth hormone therapy (Bryant et al., 2007, Naiki et al., 2013).

In a critical review of the research into psychosocial functioning in children and adolescents with idiopathic short stature, Balen et al. (2006) found most notably that these children had more psychosocial problems (social anxiety, depression, low self-esteem, poor quality of life, poorer family relationships) compared with children of normal height. Adolescence was the most challenging time where participants wanted to be like everyone else, but they found mixed results about marriage and employment prospects. This review condensed the results from eleven mainly prospective studies, however the authors did not report on the quality of these studies. These results have been replicated in a study of three hundred and forty-five children and adolescents with idiopathic short stature aged eight to eighteen from seven European countries by
Quitmann et al. (2016b). Health-related quality of life (physical and mental well-being, moods, emotions, social support) was impaired in those with short-stature compared to those who had achieved their target height.

Many studies on short stature and psychological functioning are based on parent reports, not directly from children and adolescents themselves (Gordon et al., 1982, Sandberg et al., 1994, Sandberg and Voss, 2002, Zlotkin and Varma, 2006), which Balen et al. (2006) also acknowledged in their critical review. A systematic review found that parents could not reliably rate their children’s quality of life when rating non-observable features such as psychological functioning (Eiser and Morse, 2001). There is also limited consistency of study design, methodology and outcome measures in research on psychosocial adjustment to short stature (Noeker et al., 2012), making it difficult to compare studies. Though Quitmann et al. (2016a) developed a condition specific health-related quality of life measure that has strong agreement between patent and parent ratings. The Wessex growth study was longitudinal, so therefore gives a unique insight into changes over time and was not reliant on parent reports. It is possible a larger effect would be found in adolescence as this is a difficult time and being shorter may exacerbate this (Balen et al., 2006, Claessens et al., 2005). However, the findings from idiopathic short stature compared with those of studies with “short but otherwise normal” children (Voss, 1999, pp. 370) indicate that the medicalisation of short stature may contribute to psychosocial problems (Naiki et al., 2013, Voss, 1999). For example, shorter children treated with growth hormone therapy and their parents - in a sample of children with idiopathic short stature – reported greater anxiety regarding their height compared to those not receiving treatment (Naiki et al., 2013).

1.4 Achondroplasia, Turner syndrome and Constitutional growth delay
The literature on “short but otherwise normal” children (Voss, 1999, pp. 370) shows that they are at no disadvantage compared with peers within an average height range, and those with idiopathic short stature experience significantly more psychosocial issues. What does the literature reveal about people with short stature that also have a diagnosed health condition?
Achondroplasia is a type of skeletal abnormality, which results in disproportionate short stature (Baujat et al., 2008). Gollust et al. (2003) identified that medical issues in Achondroplasia were well documented, but the ways in which the condition affected the lived experience were not, and they used surveys to explore how perceptions of Achondroplasia and quality of life differed between people with the condition and their first-degree relatives. They found that those with Achondroplasia had lower quality of life and lower self-esteem compared with their relatives, though they perceived their condition to be less serious than relatives reported. The authors acknowledged that participants with Achondroplasia “differ noticeably in appearance” (Gollust et al., 2003, pp. 454), but did not relate that to the literature on visible difference or to the outcomes of the survey. Subsequent studies report similar findings, though also lack detail regarding the lived experience. Compared to ‘healthy’ controls, participants with Achondroplasia reported persistent stature-related negative experiences (Nishimura and Hanaki, 2014). Negative appraisal style was found to mediate the relationship between these negative experiences and psychosocial issues, though no difference was found between affected participants and healthy controls in psychosocial functioning. Sommer et al. (2016) found that individuals with Achondroplasia reported body image concerns and negative experiences with strangers (staring and teasing).

Similarly, the literature regarding Turner syndrome sheds little light on what it is like to live with this condition (Sutton et al., 2005). Turner syndrome is characterised by short stature and infertility (Ranke and Saenger, 2001). In qualitative studies researchers have found that participants were most concerned about infertility, resulting in fear of initiating romantic relationships and ending up without a partner (Clauson et al., 2012, Sutton et al., 2005). Participant’s height resulted in functional and social frustrations, as they were tired of asking for help when they could not reach something or had to take public transport because they could not drive (Sutton et al., 2005). Teasing in school about being small, which for some persisted into adulthood, also troubled them (Sutton et al., 2005). Women with Turner syndrome were also found to have a higher incidence of depression compared with the general population (Sutton et al., 2005), to experience anxiety (Clauson et al., 2012) and to have lower self-esteem and negative body image compared to ‘healthy controls’ (Cragg and Lafreniere, 2010). Clauson et al. (2012) also
reported participant comparing their appearance unfavourably to unaffected siblings and peers.

The very limited research regarding psychosocial issue for people who have Constitutional Growth Delay - a condition resulting in a delay in skeletal growth and onset of puberty (Soliman and Sanctis, 2012) - has produced mixed results. Crowne et al. (1990) reviewed the growth and wellbeing of men with untreated Constitutional Delay in Growth and Puberty, which results in short stature. This case-control study compared a cohort with Constitutional Delay in Growth and Puberty to a control group of men of the same age and socio-economic status within the average height range and no diagnosed health condition. They measured self-esteem, attitudes towards height, social and employment history and found no significant difference, although over half of the sample felt that their delayed growth affected them adversely at school and socially. This study was of moderate quality; it is not clear if outcome assessors were aware of the exposure status of participants or whether participants were aware of the research question; and there is no clarity as to whether the measures were valid or reliable. It may have also been useful to compare treated and untreated participants to explore effects of growth hormone therapy. Social problems were also identified in a study with 45 adults with Constitutional Growth Delay and 980 with patients who were deficient in growth hormone (Sartorio et al., 1990). Both groups were similarly affected by their growth conditions, even though final height was better for those with Constitutional Growth Delay. It was reported that both groups experienced social isolation, were mostly unmarried and lived with their families. This quantitative study had several limitations: the findings lacked generalisability as the Constitutional Growth Delay sample was small; there was a poor response rate to the questionnaire; and it would have been useful to have general population data as a comparison for both groups. However, psychosocial issues are prevalent enough for psychosocial assessment to be recommended in these patients (Mobbs, 2005).

In summary, research suggests that people with idiopathic short stature, Achondroplasia, Turner syndrome and Constitutional Growth Delay are more likely to experience psychosocial issues (lower quality of life, depression, low self-esteem, difficulty forming relationships, social isolation) compared with people within the average height range and
no diagnosed health condition. Whether this is due to short stature or having a health condition, or both, is not easily understood. The one characteristic people with these conditions share is they look different from others.

1.5 Models and theories in chronic illness

Whilst exploring models and theoretical constructs to aid in the understanding of the lived experience of RSS as a chronic health condition it became clear that no one model or theory adequately fit the issues under investigation. This may be due in part to RSS not fitting the traditional definition of chronic illness. The (World Health Organisation, 2017) define a chronic illness as a noncommunicable disease that had a long and slow progression. The examples they give are: heart attack, stroke, cancer, diabetes, and asthma. This issue became clearer when I started to look at concepts that aimed to understand the lived experience of chronic health conditions.

Michael Bury (1982) purported that patients’ experiences of chronic illness could be understood by demonstrating how developing a chronic illness is perceived as a huge disruptive experience. The patient develops a growing dependency on those around them and are forced to reconsider plans for the future. The particular feature of chronic illness Bury described was its “insidious onset” (Bury, 1982, pp. 170) the development of the condition reaches a ‘tipping point’ that begins to disrupt the persons’ life. Williams (2000) questioned the usefulness of Bury’s theory in understanding the experience of chronic illness as it describes a typical experience of illness onset in middle to late adulthood and neglects to explain illness from birth or that developed in childhood. Williams critique is of particular significance as it exemplifies my reasons for discounting biographic disruption as a useful model to explain the lived experience of RSS. This is a congenital condition that – according to the literature – remains stable over time with no progression from ‘healthy’ to unhealthy’, as described by Bury.

Building on Williams critique (Larsson and Grassman, 2012) criticised the main characteristics of the biographical disruption model based on interviews with people who have experiences of chronic illness from childhood. They state that biographical disruption as a single event does not apply to these populations of chronically ill participants and that their experiences reflect repeated incidences of disruption across
the life span. Larsson’s critique may provide insight to understanding the experience of illness from a person with RSS perspective as it may be that biographical disruption is experienced multiple times during childhood as diagnosis, feeding difficulties, and treatments are encountered as the child develops. Although again this does not fit with RSS as these developments are not a result of an illness progressing.

1.6 Visible difference and appearance-related concerns
What one person might consider a visible difference may not be considered a difference by another, but broadly speaking a visible difference is when a person’s appearance differs from what is socially or culturally defined as the ‘norm’ (Harcourt and Rumsey, 2008). Partridge (1997) defined visible difference as ‘being significantly visibly different compared with a norm, a norm that is either defined for us by the culture or by ourselves’ (pp. 4). The appearance literature covers a wide range of visible differences such as craniofacial (Eiserman, 2001), burns (Corry et al., 2009), vitiligo (Thompson et al., 2002), facial disfigurement (Halioua et al., 2011), amputation (Mathias and Harcourt, 2014) and cancer (Williamson et al., 2010). Though, research shows that people with congenital conditions that alter appearance have similar experiences (Feragen, 2012).

People who are rated as ‘attractive’ are perceived as more friendly and helpful to others than those rated as ‘unattractive’ (Dion, 1973, Lemay et al., 2010) and people are more likely to attribute qualities such as generosity, kindness, caring, supportiveness and warmth to attractive friends and romantic partners than to unattractive ones (Lemay et al., 2010). Attractive people are also perceived as having higher morality than unattractive people (Tsukiura and Cabeza, 2011). Although there has been some criticism levelled at the attractiveness stereotype outlined above. In a review of the literature Eagly et al. (1991) found that the relationships between positive characteristics and attractiveness were not as strong as previously reported, however there was a relationship, albeit a moderate one. People perceive someone with a facial disfigurement less favourably than someone without and are significantly more likely to rate them as less capable, less effective, dishonest, untrustworthy, unemployable, and less intelligent (Halioua et al., 2011). Goffman (1963) proposed that ‘undesirable’ characteristics may lead us to no longer see that person as a ‘fellow human being’, but reduced to a label society has given them, and if that identity or group is not to our liking or the norms
society has created, we may see that person as suboptimal. This suboptimal attribute is a stigma (ibid).

Society has manifested itself in such a way that is conducive for people to form groups where some characteristics are valued over others when forming a membership to these groups (Goffman, 1963). Upon a first encounter with a person we most likely attribute them to a group and make assumptions as to what that person might be like. Scambler and Hopkins (1986) built on Goffman’s work by introducing the concepts of enacted and felt stigma. They described enacted stigma as actual acts of discrimination against a person who is perceived as suboptimal or possessing characteristics not valued by society. Felt stigma, a more complicated phenomenon, is the fear of being stigmatised and feelings of shame around the potential stigmatising characteristic or behaviour. Felt stigma can lead to behaviours such as non-disclosure and attempts to ‘fit in’. Nevertheless, the concept of stigma has been challenged on the count of a failure to address more distal issues. Fine and Asch (1988) criticised the focus on how stigma affected the person rather than societal factors that lead to the label in the first place. Link and Phelan (2001) criticise research that has been conducted using the concept of stigma. They observed that most researchers study this concept from the vantage point of the un-stigmatised and use frameworks to understand stigma not situated in the lived experience of the group for which they are applying the framework.

As I have previously outlined, people who have a visible difference or who are perceived as being ‘unattractive’ may be attributed certain undesirable characteristics. These attributions may lead to discrimination and stigmatisation, even by peers and family members, which may put a persons’ wellbeing at risk (Rumsey et al., 2004) Importantly, who will or will not experience psychological distress cannot be predicted based on objective measures of how severe the visible difference appears to others (Rumsey et al., 2004); experiences of stigma and strategies such as social comparison have an impact on adjustment (Galioto and Crowther, 2013, Krayzer et al., 2008, Taylor, 1983). If psychological distress is also based on the person’s own appraisal of their difference, psychosocial factors need to be explored. What follows is a description of factors that may contribute to maladaptive adjustment and factors that may result in positive adjustment to looking different.
Being visibly different or having appearance-related concerns can impact significantly on an individual resulting in psychosocial problems such as: anxiety (Griffiths et al., 2012, Rumsey et al., 2004, Williamson, 2014); depression (McBain et al., 2013, Rumsey et al., 2004, Stock et al., 2015, Williamson, 2014); low self-esteem (Griffiths et al., 2012, Lovegrove and Rumsey, 2005, Stock et al., 2015, Williamson, 2014, Williamson et al., 2010); social anxiety; and social avoidance (McBain et al., 2013, Rumsey et al., 2004, Williamson, 2014, Williamson et al., 2010). A visible difference can also influence how others treat that individual: bullying, teasing and ridicule are sometimes reported by children (Buhlmann et al., 2011, Lovegrove and Rumsey, 2005, Griffiths et al., 2012, Almenara and Jezek, 2015, Magin, 2008) and is often centred around body and facial appearance (de Oliveira, 2015) and for those with a visible difference a minority can experience significant bullying causing psychological distress (Magin, 2008). This can result in low self-esteem in adolescence (Griffiths et al., 2012, Stock et al., 2015, Williamson, 2014, Williamson et al., 2010) and extend into adulthood (Rumsey and Harcourt, 2005b).

Comments from others, bullying and teasing may serve to reinforce the individuals’ negative appraisal of their appearance and in turn increases social anxiety and decreases self-esteem and confidence making it more challenging to build friendships, forge romantic relationships (Griffiths et al., 2012, Mathias and Harcourt, 2014) and embark on endeavours such as studying or building a career (Stone and Wright, 2013). At the heart of social anxiety is worry about how others perceive us, and the way we look and/or behave, resulting in a loss of ‘social capital’ and ultimately being shunned (Clark and Wells, 1995). Social capital describes how engaging effectively with others or having status, for example, ‘social attractiveness’ (Gilbert, 1997, pp. 115) results in an increase in connections and access to others (Lin, 2001). This in turn secures benefits such as reaching goals, increased psychological wellbeing, and better opportunities (employment for example) (Lin, 2001). Social capital, social attractiveness and status drive processes such as social comparison and self-criticism (Gilbert, 1997).

A person may feel rejected by others because of their appearance, but poor social skills due to the avoidance of social situations may exacerbate exclusion and loss of social
capital (Kent, 2000). Social situations may lead to physiological changes such as blushing, sweating and rapid heartbeat or the person may appear less congenial as they are preoccupied with how they are feeling; people around them may not act as positively towards them leading to a confirmation that they are not liked or valued (Kent, 2000), creating a downwards spiral. Ultimately this could lead to avoidance of certain situations or social encounters altogether. Reduced social contact can impact significantly on feeling of connectedness, job prospects and finding or retaining romantic relationships (ibid). Positive emotion and psychological wellbeing result from forming and maintaining relationships, whereas any threat, even potential threat to these relationships can lead to psychological distress (Baumeister and Leary, 1995).

On the other hand, many people seem to cope effectively with the challenges of looking different (Noeker et al., 2012, Vilhjalmsson et al., 2012). For example, Williamson et al. (2010) reported that adolescents and parents managed the psychosocial impact of appearance change during cancer treatment by using humour and over confidence to ameliorate social awkwardness experienced by them and those around them. Positive adjustment to adversity is often termed as being ‘resilient’. Resilience has been defined for the purposes of research as “a dynamic process encompassing positive adaptation within the context of significant adversity” (Luthar et al., 2000, pp. 513) or “successful adaptation or the absence of a pathological outcome following exposure to stressful or potentially traumatic life events or life circumstances” (Seery et al., 2010, pp. 1025) and in the public domain as “our ability to cope with and bounce back from adversity” (Action for Happiness, 2016).

Skills such as resilience and coping are important, and as I have outlined above, negative emotions and poor adjustment are not just related to reactions from others, but to how that person perceives and copes with this visible difference (Rumsey and Harcourt, 2012). Feragen et al. (2010) compared young people with a visible cleft lip/palate to a control group of young people with a non-visible cleft and found the control group to have a higher incidence of depression and lower scores on functioning overall. The authors concluded that this was due to a higher emotional resilience among the visible difference sample. This could also be due to a higher exposure to adversity resulting in hardiness to adversity (Rutter, 2009), which suggests acceptance of the situation rather than a positive
adaption. Schanke and Thorsen (2015) interviewed ten people with short stature aged between 45-65 exploring stigma handling, and found that avoiding being stigmatised and staying resilient was necessary throughout the lifetime to avoid negative effects such as depression and anxiety. In addition to these findings, Masten et al. (1999) found the more adversity a child encountered growing up, the less emotional resources they are likely to have to remain resilient.

Research in the field of appearance and visible difference uses a mixture of approaches and study designs and reports on a variety of conditions (cleft lip/palate, cancer, rheumatoid arthritis, skin conditions, burns, amputation). A substantial amount of studies were conducted in the UK, so are culturally and socially relevant to this study, and most have equal gender representation. There are fewer studies that focus on appearance-related concerns in adults, as many have studied these issues in an adolescent population. There is still an underrepresentation of research exploring appearance-related concerns in men (Parent et al., 2016, Thompson, 2012), and how being visibly different impacts romantic and intimate relationships is currently under-explored (Griffiths et al., 2012). It may also be the case that this research under reports the impact that visible differences has; for example, one study reported that parents declined to allow their child to participate in research on appearance due to a worry it may heighten a pre-existing concern (Williamson et al., 2010).

1.7 Conclusion
The issues experienced by people with idiopathic short stature, Achondroplasia, Turner syndrome and Constitutional Growth Delay are very similar to the experiences of people with a visible difference. As with any syndromic condition there are many characteristics that may affect the individual in various ways. With RSS, height is only one characteristic, there are other characteristics that may result in that person feeling or being visibly different. There is a lack of research exploring the lived experience of RSS, and a clear need for this to aid understanding. The Child Growth Foundation, the UK’s leading charity dedicated to childhood growth disorders, has indicated to researchers that information on what it is like to live with RSS in adulthood is a major priority area for research. Families in touch with the Child Growth Foundation have requested as much information as possible above and beyond physical health, to aid in the decision making process for
RSS treatment. Information beyond childhood and adolescents about the lived experience of those that have and have not undergone treatment will make that decision for families a more considered one. This research documents what it is like to live with RSS in a sample of 15 adults, furthering the understanding for health professionals treating patients with RSS, and patients living with RSS and their families. It provides a much-needed perspective on the future for patients with this rare genetic syndrome.

1.8 Aim
To explore the lived experience of people with RSS.

1.9 Research questions
RQ1. What are participants’ perceptions of having RSS?

RQ2. Are there challenges in adulthood of living with RSS? And if so, what are those challenges?

RQ3. How does having RSS change over time? Are there time points where healthcare could be targeted?

RQ4. What do patients, families and health professionals need to know about living with RSS?
2.0 Methodology

The sections that follow describe the methodology and method of a qualitative study that ran concurrently with a quantitative study, both investigating Russell-Silver syndrome. The quantitative study recruited 34 participants and aimed to investigate the effectiveness of growth hormone therapy on height in people with RSS and health in adulthood.

Methodology refers to the way in which we obtain knowledge (Braun and Clarke, 2013). The methodology used in this study was underpinned by decisions I made about how best to obtain knowledge, as close to reality as possible, regarding living with RSS. What counts as meaningful knowledge is informed by ontological and epistemological assumptions; table 2 below defines ontology and epistemology with various positions listed beneath.
Table 2: Ontologies and epistemologies in psychology (adapted from Braun and Clarke (2013)).

<table>
<thead>
<tr>
<th>Ontology</th>
<th>What constitutes reality; how do we interpret and understand the world around us?</th>
<th>Epistemology</th>
<th>What counts as valid knowledge; what is legitimate knowledge and how can we obtain it?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Realism</td>
<td>What is real is independent of human ways of knowing it and there is only one version of that truth. With the correct methods you could truthfully observe reality</td>
<td>Objective</td>
<td>Positivism</td>
</tr>
<tr>
<td>Relativism</td>
<td>What is real depends on how humans interpret reality, it is entirely constructed and there are multiple truths. What is real depends on time and context</td>
<td>Subjective</td>
<td>Constructionism</td>
</tr>
<tr>
<td>Critical realism</td>
<td>There is a reality to know, but it could only be known by examining layers of subjective realities</td>
<td>Strives for objectiveness</td>
<td>Contextualism</td>
</tr>
</tbody>
</table>
This was the first time in my academic career I had encountered these philosophical concepts and, on reflection, it was a steep learning curve. I found it an effortful process, not only understanding the concepts, but also taking the next step in appreciating how they apply to my research and how they influence decisions I made about answering the research questions. I made the connection between deciding what I, the researcher, consider to be real and legitimate knowledge, how I should garner that knowledge to produce a ‘good’ piece of research. As Kelly (2009) said, attending to theoretical issues throughout the research process and explicitly stating theoretical positions is indicative of quality in research as it allows consistency to be applied throughout the process.

I conducted this research based on the assumption that there is a reality to know and we can know the lived experience of RSS, but this reality is multi-layered (Alvesson and Skoldberg, 2009). The way in which individuals attach meaning to experiences and how broader social constructs, such as body image or social comparison, interact with that meaning making (Braun and Clarke, 2006, Braun and Clarke, 2013) contribute to this rich multi-layering of reality. By collecting participant experiences, I could strive to come to a truth whilst acknowledging that knowledge is dependent on context and interpretation (mine and my participant’s) (Madill et al., 2000). I believed that each participant’s experience of living with this condition, although seen through a lens coloured by their own beliefs and interpretations, is true for them (Madill et al., 2000). If I had taken a more relativist or constructionist view, whereby objectivity is not possible, there would be no opportunity to make wider claims from the data to inform future practice and intervention. Therefore my philosophical and theoretical decisions were based on my beliefs about what was real and valid knowledge, but my decisions were also grounded in pragmatism and by what was expected by stakeholders who required applicable knowledge.

2.1 Qualitative methodology
Qualitative researchers are interested in the meaning and complexity of a subject and exploring phenomena that cannot be adequately explained numerically; a qualitative methodology is best placed to understand what it is like to live with a particular condition or to have experienced a certain phenomenon (Braun and Clarke, 2013, Kelly, 2009,
Willig, 2013). I chose qualitative methodology for two main reasons. Firstly, I felt qualitative methods would bring the patient voice to complement the objective literature, providing a fuller picture. And secondly, I believed there to be multiple interpretations of the phenomena under investigation.

2.2 Data generation
Two possible methods of data generation were considered for this study: focus groups, and in-depth, semi-structured interviews. Observational methods and diaries were not appropriate, as both would show a snapshot of what it was like to live with the condition and not elicit detailed experience across the lifespan. Focus groups may have been an appropriate method to explore the collective experience of the condition and are not so time intensive, but I could not have explored the area in as much depth and participants may not have been comfortable discussing sensitive issues in a group. Also, for pragmatic reasons it would not have been possible to organise participants from all over the UK, as this condition is rare, to come together in one location.

I decided upon using in-depth, semi-structured interviews for the following reasons. Firstly, they are the most frequently used qualitative method of data generation in psychological research and compatible with many methods of analysis (Willig, 2013). Secondly, they focus on drawing out individual experiences and allow the researcher to explore specific concerns or issues, to be flexible about the wording of questions, and add questions about relevant topics that arise (ibid). Thirdly, they are well suited to generating data for experience-type research questions (Braun and Clarke, 2013). Fourthly, validity is enhanced when using semi-structured interviews as the questions can be tailored and participants have the opportunity to discuss issues relevant to them and their health condition (Mason, 2002). Lastly, semi-structured interviews allow the researcher to gather in-depth information for one participant over an extended period of time (Harding, 2013). The main disadvantage of using interviews over focus groups was the time it takes to conduct and transcribe them (ibid).

2.3 Method of analysis
Based on the research aim, I considered interpretive phenomenological analysis (IPA) and thematic analysis (TA) as possible methods of analysis. Both were suitable for exploring
the lived experience (Clarke et al., 2015, Smith and Osborn, 2008). TA was chosen over IPA for several reasons. As a researcher new to qualitative methods, TA was a good method to start with, as it develops the skills needed for most other forms of qualitative analysis (Braun and Clarke, 2006) and methods such as IPA are more challenging for the novice researcher. TA is a flexible method of analysis (ibid) and can be used with a wide range of research questions, larger sample sizes than other methods such as IPA, and the results are easier for a wider audience to interpret i.e. parents of children with RSS (ibid). TA also allows the researcher the capacity to make interpretations of how participants make sense of the world, whereas IPA makes the assumption that participants are able to communicate to the researcher “the subtleties and nuances of their physical and emotional experiences” (Willig, 2013, pp. 95) Using TA allows the research to draw on psychological concepts to explain participant experiences. The decision to use TA was also informed by the epistemological assumptions, as it is not aligned to any particular theoretical framework (ibid). The knowledge TA generates, in the form of themes that describe how people experience a particular phenomena, corresponded well with the research questions.

There are many methods of TA (Aronson, 1994, Boyatziz et al., 1998, Joffe, 2012); I used the Braun and Clarke method (see table 1 for key stages of analysis) primarily for pragmatic reasons, as it was the method taught on my professional doctorate and I had become familiar with it. And secondly, the resources to support the method gave detailed, step-by-step instructions, which enabled me as a researcher new to qualitative methods to feel confident in my analysis. Braun and Clarke describe TA is “a method for identifying, analysing and reporting patterns (themes) within data” (Braun and Clarke, 2006, pp. 79); Grbich (2013) describes it more generally as a popular method of data reduction. Although I used a particular method of TA, Grbich (2013) makes the case that there are no set definitions for codes and themes and no set rules for the analysis of qualitative data, but the important aim is to clearly define how you have used the terms and to be as transparent as possible when writing up the process of analysis. Another criticism of TA and also of coding and analysing qualitative data is that the researcher takes out concepts that are rooted in the participant’s life and how they make sense of their world, and then makes their own interpretations, taking the process a few steps away from reality (Holloway and Jefferson, 2000). And, as Joffe and Yardley (2004) argue,
maybe this is not a problem as TA identifies themes across a group of people, has a cross-sectional aim, and is not there to tell the story of individuals. However, I did consider the possibility of constructing vignettes as an additional form of representing my data, which would provide a detailed story for one or two participants. This was not possible due to the length of the project and time constraints.

Figure 2 shows the basic steps taken when conducting a TA. I have illustrated this as circular and included ‘two-way’ arrows between phases to highlight that TA is not a linear process, whereby a researcher completes one phase and moves onto the next, it is very much a ‘back-and-forth’ process. Phase 1 involves immersing yourself with the breadth and depth of the data, repeated reading of data, note taking and generating a list of ideas. In phase 2 the researcher works systematically through the entire data set, producing initial codes and identifying interesting aspects that may form the basis of repeated patterns (themes). Phase 3 requires the sorting of codes into themes, visual representations could be used to start organisation, and start thinking about relationships between themes and sub-themes. In phase 4 themes are refined (some may not have had...
enough data to support them, some may be split and some may be combined), codes checked under each theme, and themes are checked to ensure they reflect the entire data set. The researcher should know what the themes are, how they fit together and what story they tell. By phase 5 the researcher should have a thematic map and will be refining and defining themes, writing a detailed analysis for each theme, and thinking of theme names for the final analysis. By phase 6 the researcher will be able to tell the complicated story of their data by including data extracts to illustrate the story. The analysis must go beyond what the data says and argue how it answers the research questions.

**2.4 Researcher influence on data generation and analysis**

When using a qualitative approach the researcher is required to think about how their experiences, beliefs and attitudes affect the data generation and analysis. For this reason I kept a research diary throughout the duration of this project (see appendix 1 for an extract), which was recommended good practice to demonstrate transparency and reflexivity (Braun and Clarke, 2013). This aided in the development of the research and contributed to a rich and complex analysis of the data. In this journal I reflected on decisions made with analysis, problems overcome, observations made when interviewing, emotions felt and anything else that seemed pertinent. My experiences, beliefs and attitudes may have influenced the data generation and analysis in the following ways:

1. I did not have RSS, know a person with RSS, or have a diagnosed genetic or health condition. It may be common for people to research areas in which they have a personal interest or investment. This comes with the advantages of having an in-depth knowledge of the unique challenges and also gaining participants trust. Although, not having any personal investment or previous views and attitudes about RSS meant I had no fixed agenda about what I wanted to discover in the data.

2. I was a health psychologist in training, not a medical doctor. I feel that this was particularly salient given the dearth of psychosocial research about people with RSS. Viewing RSS through a psychosocial lens allowed me to go beyond the literature, something a medical doctor may have struggled to do. Although, there
is the possibility that I did not identified significant themes in the data that a sociologist or a medical doctor would have.

3. Alongside working as a researcher and interviewing people with RSS I was also a smoking cessation advisor. This brought advantages such as confidence in meeting new people, in dealing with emotive discussions and motivational interviewing skills, which help to open up discussion. The disadvantage being I was more familiar with helping people solve problems through action planning and other techniques. This was not appropriate in an interview as I was there to listen and document. This passive style felt frustrating at first, as I had not anticipated how this change in dynamic would make me feel. Once I had reflected on this and my role as an interviewer, it became easier.

4. I was a 39 year-old woman interviewing males and females. I did ask some personal questions about romantic relationships and participants may have found that a challenging topic to discuss with someone of the opposite sex. Therefore some participants may not have been as open with me as they would if I were a man.
3.0 Method

3.1 Participants

I interviewed 15 participants, 6 were female and 9 were treated with growth hormone (GH). The participants’ ages ranged from 25-69 with a mean of 38. No participants were currently receiving specialist medical support for RSS and were recruited to the quantitative study researcher by a range of methods including dissemination of study information through support groups, such as the Child Growth Foundation, and by liaising with genetic centres across the UK. See table 3 for demographic information.

Table 3. Demographic information

<table>
<thead>
<tr>
<th>Men*</th>
<th>Age range</th>
<th>Growth hormone</th>
<th>Height</th>
</tr>
</thead>
<tbody>
<tr>
<td>Darin</td>
<td>31-40</td>
<td>✓</td>
<td>5’</td>
</tr>
<tr>
<td>Todd</td>
<td>31-40</td>
<td>x</td>
<td>5’6”</td>
</tr>
<tr>
<td>Warner</td>
<td>31-40</td>
<td>✓</td>
<td>4’9”</td>
</tr>
<tr>
<td>Oli</td>
<td>31-40</td>
<td>x</td>
<td>5’1”</td>
</tr>
<tr>
<td>Grant</td>
<td>21-30</td>
<td>✓</td>
<td>5’6”</td>
</tr>
<tr>
<td>Eric</td>
<td>31-40</td>
<td>✓</td>
<td>5’6”</td>
</tr>
<tr>
<td>Elliot</td>
<td>31-40</td>
<td>✓</td>
<td>5’5”</td>
</tr>
<tr>
<td>Luke</td>
<td>31-40</td>
<td>✓</td>
<td>4’8”</td>
</tr>
<tr>
<td>Glenn</td>
<td>61-70</td>
<td>x</td>
<td>5’</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Women*</th>
<th>Age range</th>
<th>Growth hormone</th>
<th>Height</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bonnie</td>
<td>31-40</td>
<td>x</td>
<td>4’10”</td>
</tr>
<tr>
<td>Maddy</td>
<td>21-30</td>
<td>✓</td>
<td>5’3”</td>
</tr>
<tr>
<td>Judy</td>
<td>31-40</td>
<td>x</td>
<td>4’3”</td>
</tr>
<tr>
<td>Mina</td>
<td>21-30</td>
<td>✓</td>
<td>4’8”</td>
</tr>
<tr>
<td>Wendy</td>
<td>41-50</td>
<td>x</td>
<td>4’7”</td>
</tr>
<tr>
<td>Joanne</td>
<td>51-60</td>
<td>✓</td>
<td>4’9”</td>
</tr>
</tbody>
</table>

*Average height for men in the general population is 5’8” (Royal College of Paediatrics and Child Health, 2015) compared with 5’2” in this sample of men with RSS. Average height for men in this sample who received growth hormone treatment was 5’1” and 5’2” for those who received no treatment.

*Average height for women in the general population is 5’4” (Royal College of Paediatrics and Child Health, 2015) compared with 4’8” in this sample of women with RSS. Average height for women in this sample who received growth hormone treatment was 4’9” and 4’5” for those who received no treatment.

The quantitative arm of the wider study recruited 34 participants with RSS and I recruited participants from this sample. My aim was to recruit approximately 15 participants; this number was decided upon through researching the literature on sample size in qualitative research, by considering methods such as saturation, the point in analysis where no new ideas are generated by the data (Braun and Clarke, 2013), and by constraints such as the rareness of the syndrome and the word count of a professional doctorate thesis.

2 All participant names are pseudonyms.
compared with a PhD thesis. Using saturation as a method of informing sample size may never be possible as each new participant will offer something new to add to the data (Josselson and Lieblich, 2003). It can be argued that ‘saturation’ is rarely achieved. In a sample of 560 PhD studies using qualitative interviews it was found that the average sample size was between 20 and 30 (Mason, 2002). Most sample sizes were multiples of 10 from which the author concluded there was no logical explanation and clearly demonstrated that ‘saturation’ as a method of determining sample size was not being used. Josselson and Lieblich theorised that a more pragmatic approach, whereby it is more likely the researcher becomes saturated and this is what dictates sample size. This best describes how I arrived at the final number of participants needed for this study. Interviewing people with RSS generated detailed and rich data; this in addition to time constraints to complete my thesis informed the final number of participants.

Initially I adopted a purposeful sampling strategy (Patton, 1990), which involved deliberate sampling to aid the purpose of the research (Harding, 2013). I included participants across a range of ages, treatments undergone and gender. A broad spread of ages allowed the capture of different experiences over time and a range of participants who had undergone different treatment options, such as growth hormone, allowed me to search for similarities and differences between participants during the analysis. As the study progressed this sampling strategy became redundant, as recruitment was slow to the quantitative arm of the study. This meant that I contacted all participants from this arm who consented to participate in future research and met the inclusion criteria, resulting in all those who responded being interviewed.

The main inclusion/exclusion was genetic conformation of diagnosis and age. Only participants with a genetic confirmation of RSS were interviewed, rather than diagnosed on the basis of clinical characteristics alone (see ‘Ethical Dilemmas’ below for more detail). Participants aged 18 and over were recruited to this study to obtain a more homogeneous sample for analysis.

3.2 Recruitment strategy

The researcher for the quantitative arm asked for an expression of interest during the consent process for taking part in future research; I contacted only those who agreed to
this (all participants consented to this). Participants were contacted by post initially and were sent a cover letter (appendix 2) with an information sheet (appendix 3), which included details of how to take part in the study. If after a few weeks, I had not received any correspondence I emailed the same information and then followed this up with a telephone call. After three methods of contact I did not feel it was appropriate to persevere.

Nineteen potential participants from the wider study were identified as fitting the inclusion/exclusion criteria. Figure 3 describes how 15 participants were recruited.

![Figure 3. Diagram of recruitment](image)

3.3 Interview schedule
The interview schedule (see appendix 4) was constructed to cover the key stages of peoples’ lives, for example education, work, friends, family, intimate relationships and to address how RSS has impacted these areas and whether living with RSS had changed over time. It was ordered according to significant events in people’s lives, so it started with any early memories of having the condition, then moved onto school, college etc. Researching the RSS literature, literature on similar conditions such as Achondroplasia and Turner
syndrome, and sitting in on the medical appointments for the quantitative study also informed the schedule.

I piloted the interview schedule with a work colleague who had a chronic health condition to assess how appropriate the questions were, if they were in an order that made sense and also to have some practice in asking the questions. I made several adjustments to the schedule (see appendix 5 for tracked changes of interview schedule); these were mainly taking out questions that were difficult to answer, taking out questions that repeated themselves and reordering questions. The interview schedule continued to evolve as I progressed through the interviews and as developing themes were identified in the data.

I had met 5 participants previously at their appointment for the quantitative arm of the research. This was arranged for two reasons: firstly at the beginning of the research it gave me the opportunity to meet people with the syndrome I was studying, which then helped inform the interview schedule by highlighting areas of life that the syndrome affected. And secondly it gave participants the opportunity to meet me first to help build rapport before asking if they would like to take part in the in-depth interview stage of the research.

3.4 Procedure

3.4.1 Consent
All participants were provided with an information sheet and insured they had the opportunity to discuss the research and ask questions before they agreed to be interviewed. I gave them a consent form to read and sign before the interview started. I reassured the participant that we could stop the interview at any time if they wished and that it would be audio recorded, but this could be turned off at their request. I then summarised the aim of the study before I turned the dictaphone on and commenced the interview.

I explained to participants I was not trained in medicine and that I had a background in psychology before the interview commenced to ensure they had no expectation I would be able to answer any medical questions they had. This was an issue other qualitative
researchers in the research group I work with had experienced, as participants had asked them questions and wanted medical advice they could not provide. So by making my qualifications and background clear to participants I did not experience these issues. On some occasions I sought details of support services if issues arose in the interview. For example, I found the details of the local health trainer service for a participant that was anxious about being overweight.

I did not interview anyone who lacked capacity to consent to take part in my research as this was screened for in the wider study. I had to consider the issue of capacity as the literature states that learning difficulties could be a characteristic of RSS. The literature does provide information on IQ scores, which would indicate that at worst a participant could be classed as having ‘mild’ learning difficulties (British Institute of Learning Disabilities, 2011). Therefore, I believed capacity to not be an issue in this particular study. This decision was based on the literature, as well as consultations with the rest of the clinical team, which was comprised of a professor of medical genetics and a paediatrician, who both had considerable experience with this patient group. They also observed that these patients do not identify themselves as people who have a learning disability or lack capacity. Interviews were conducted between March 2015 and January 2016.

3.4.2 Debriefing
During the in-depth interview, participants discussed their life experiences. This often involved the discussion of sensitive, difficult or emotionally charged issues. Participants were offered the option to stop the discussion if they wished to. I aimed to avoid any unnecessary intrusion (Social Research Association, 2003) as some of the questions were around sensitive issues such as romantic relationships.

3.4.3 Transcription/management of data
I transcribed the first interview myself, as I had not done this task previously and I also transcribed an interview with a person who had a speech impediment as this was easier for me to transcribe as I had taken notes to aid recall. Due to having money in the budget and time constraints a transcription service was used for the remaining audio recordings. This had the advantage of being incredibly time efficient, but the potential disadvantage of removing the opportunity of familiarisation with the data, the first step in the
analytical process (Braun and Clarke, 2006). To counteract this I verified each interview after they had been transcribed, which allowed me to listen to the interview again before the transcription was finalised and to ensure the accuracy and anonymity of the data.

3.5 Data analysis

Figure 1 (Braun and Clarke method of TA) sets out clear steps in the analysis of data. The stages were helpful to me as a researcher new to qualitative methodologies and TA, however, I found the process to be much more iterative. It involved moving back and forth through the stages until I became satisfied with what I had identified and how that fitted into my conceptual map. Once the participant interviews were transcribed and verified I read through each one, writing comments along the side of the transcript (see appendix 8 for an example). I paid particular attention to pertinent elements of the data in relation to the research question, continually asked myself questions about the data, identified where participants had contradicted themselves or commented on how the participant was making sense of their experience. When a few transcripts were completed I collaborated with another member of the study team who had completed the same task to discuss our thoughts.

After this discussion, tentative codes were identified relating to the research questions or that seemed to occur frequently and were also salient. I coded the first few transcripts using NVIVO and the tentative codes. The process of commenting and then coding was repeated with a few more transcripts. This led to the adjustment of some codes by either checking the data under that code to ensure it encapsulated what the code was describing, or taking broad codes and breaking them down further to be more meaningful.

During the process of coding data and refining the codes I started to experiment with organising them into themes to identify conceptual frameworks. For example, I identified that five or six separate codes were all illustrating ways of coping.
3.6 Ethical issues

3.6.1 Confidentiality
Each participant had a study number from the quantitative arm of the research, which I also used for interview data. I transferred the digital recordings to a password-protected computer and destroyed them once transcriptions were completed and verified. Participant confidentiality was ensured in accordance with the NHS Code of Confidentiality, specifically relating to pages 7-8 which detail ‘Using Confidential Patient Information’, ‘Obligations on Individuals Working in the NHS’, ‘Consent Issues’ and ‘Data Protection Considerations’. The file linking ID numbers and participants’ names were stored on an NHS computer in a secure and separate location from the remainder of the research data. Careful consideration was given when including quotes in the writing up of this research, as identification, despite anonymisation, was possible due to the small number of participants with this rare condition.

3.6.2 Ethical dilemmas
I encountered several situations in which I sought guidance throughout the research process. The first was the participant who had a speech impediment so significant I did not think I would be able to transcribe the interview through an audio recording. My very first reactive thought was to not include him in the in-depth interviews, however I knew this would be extremely unethical, unfair and his contribution to the research could be invaluable. In collaboration with the team and seeking the advice of an expert in the field of interviewing people with cleft palate I came up with a solution that worked effectively. This solution was to record his voice, whilst also taking quite detailed notes during the interview; this allowed me to ‘anchor’ my notes to the audio recording.

The second dilemma was encountered whilst at the Child Growth Foundation conference where I met people with clinical features of RSS but with an unknown genetic cause. They were enquiring about taking part in the study and it felt unethical to exclude them from participating. After a discussion with the wider team it was decided that they would not be invited to take part in this study, as we only recruited people with a confirmed genetic diagnosis, but could be offered the opportunity to participate in the 100,000 genomes project (Genomics England, 2016). This felt like a more appropriate project for these particular patients to take part in as the rare disease arm aims to search for a diagnosis.
3.6.3 Researcher safety
A ‘buddy system’ was developed with another member of the research team in accordance with the University Hospital Southampton Foundation Trust and University of the West of England ‘Lone Worker’ policies (see appendix 9).

3.6.4 General ethics
The research was approved by the NHS REC (Reference number: 13/SC/0630) and the ethics committee at the University of the West of England (see appendix 10 for letters of confirmation).

3.7 Quality in qualitative research
Demonstrating quality, or validity, in qualitative research is not straightforward as quantitative methods have until recently, dominated psychological research (Mason, 2002, Tracy, 2010, Yardley, 2008). Terms such as reliability and generalisability are often inappropriately applied to qualitative research (Yardley, 2008, Tracy, 2010) and some assume that qualitative research should have its own terms to measure quality (Gergen, 2014, Hannes et al., 2010, Seale, 1999). Validity in research refers to the credibility of a study and reliability and generalisability help demonstrate this. In reference to reliability, argues that the qualitative researcher cannot produce tools that generate the same data again and again. This is not the nature of qualitative research where context and individual differences are of interest rather than generalisable laws (Yardley, 2008). Generalisability is not possible (although theoretical generalisability is), and maybe not even desirable, in a qualitative study. It may not be practical to generate enough rich data to be generalisable and also qualitative research is often aiming to provide insights from one small group or context that could make claims that could be applied to similar groups (Yardley, 2008). Mason (2002) suggests making wider theoretical claims for the findings of a study; for example, I could state that I had no reason to believe my sample will be atypical of the whole population of people who had RSS. I was concerned the participants that came forward to take part may have only been people who were greatly affected and wanted help or support, but I had a wide range of people who were affected in a wide range of aspects, some more severely than others. This position could also be supported by looking at the literature, which documents how RSS manifests itself and
how it impacts on those with the condition, and also by the experts in RSS who are working on the quantitative arm of the study.

I employed several techniques to ensure quality in my research. For example, to enhance the validity of the findings from this research I was as transparent as possible in regard to the analysis of the data. I gave full explanations with examples demonstrating conceptual interpretations of the data, I reflected on any decisions made and asked the opinion of other members of the team to ensure I explored perspectives other than my own (Mason, 2002). Validity could have been enhanced by gaining the opinion of participants, as they had the unique position of living with the condition I was studying, a very different position to my own (Mason, 2002), but this was not possible within the scope of this thesis. I also used Tracy (2010) ‘big-tent’ criteria (see table 4) as guidance to enhance validity and overall quality. The aim of Tracy’s eight criteria is to demonstrate quality to those who see there being a hierarchy within the different research paradigms and also as a guideline to use when designing and conducting any research in the social sciences. There are many alternative criteria to inform quality in qualitative research (Henwood and Pidgeon, 1992, Meyrick, 2006, Yardley, 2015), but I chose to use this guidance to ensure this research was to a ‘good’ enough standard because the criteria was well described, comprehensive and a good fit for this research.
<table>
<thead>
<tr>
<th>Criteria</th>
<th>Description of criteria</th>
<th>Application to this study</th>
</tr>
</thead>
<tbody>
<tr>
<td>Worthy Topic</td>
<td>Relevant, timely, significant, interesting</td>
<td>Research was requested by a patient group, there was no research in this area at the time of writing and it also questioned ‘taken-for-granted assumptions’ (pp. 840) that short stature has the biggest impact on the lives of people with RSS</td>
</tr>
<tr>
<td>Rich Rigor</td>
<td>Theoretical constructs, data and time in field, sample, context, data collection and analysis</td>
<td>I was extremely fortunate that the participants I interviewed talked candidly about their experiences allowing the collection of rich, in-depth data on the lived experience of an RSS person. I was able to identify several common themes in the data and make conceptual interpretations as well as descriptive ones</td>
</tr>
<tr>
<td>Sincerity</td>
<td>Self-reflexivity, transparency</td>
<td>I kept a research diary in which I recorded each step of the process, but also my thoughts on what I brought to the generation of data and analysis. I have been open about my inexperience in qualitative research and accepted support where necessary</td>
</tr>
<tr>
<td>Credibility</td>
<td>Thick description, tacit knowledge, triangulation, member reflections</td>
<td>I discussed any interpretations and finding with a member of the research team, who was also familiar with the transcripts from each participant</td>
</tr>
<tr>
<td>Resonance</td>
<td>Evocative representations, naturalistic generalisations, transferrable findings</td>
<td>The findings resonate with findings from other genetic conditions such as cleft palate and Turner syndrome</td>
</tr>
<tr>
<td>Significant contribution</td>
<td>Conceptually/theoretically, practically, morally, methodologically, heuristically</td>
<td>My research could give people with RSS a voice to counter what is reported in the medical literature and the disturbing pictures in medical case studies. This research will also add to the literature in other fields such as body image and appearance</td>
</tr>
<tr>
<td>Ethical</td>
<td>Procedural, situational, cultural, relational, exacting ethics</td>
<td>Attending the good clinical practice training ensured that I understood how and why the procedural aspects of the research needed to be carried out in particular ways. For example, I had never used a site file before. When all my interviews drew to a close I reflected on whether the participant was in the same state or better than I had found them. I also thought in-depth about consent (see reflection in appendix 11)</td>
</tr>
<tr>
<td>---</td>
<td>---</td>
<td>---</td>
</tr>
<tr>
<td>Meaningful coherence</td>
<td>Achieves what it purports to be about, uses appropriate methods, meaningfully interconnects literature, research questions, findings and interpretations with each other</td>
<td>I gave a considerable amount of thought to how this research was designed to ensure I was clear about my aims and that my design and measures matched that aim. My method of analysis also corresponded well with my skills as a new researcher</td>
</tr>
</tbody>
</table>
4.0 Findings

Three themes were identified during the analysis of this data. “It’s not just all about height” highlighted participants’ appearance-related concerns, which stemmed from differences extending beyond short stature to issues with asymmetry, weight, and speech to name a few. ‘Resilience – threat and preservation’ described adversity and the ways in which participants coped. And lastly, “Mayor of the friend zone” illustrated how participants dealt with their visible difference; male participants in particular experienced negative consequences psychologically and romantically due to their visible differences.

Table 5. Participant age range, height and treatment.

<table>
<thead>
<tr>
<th></th>
<th>Age range</th>
<th>Growth hormone</th>
<th>Height</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Men</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Darin</td>
<td>31-40</td>
<td>✓</td>
<td>5’</td>
</tr>
<tr>
<td>Todd</td>
<td>31-40</td>
<td>X</td>
<td>5’6”</td>
</tr>
<tr>
<td>Warner</td>
<td>31-40</td>
<td>✓</td>
<td>4’9”</td>
</tr>
<tr>
<td>Oli</td>
<td>31-40</td>
<td>X</td>
<td>5’1”</td>
</tr>
<tr>
<td>Grant</td>
<td>21-30</td>
<td>✓</td>
<td>5’6”</td>
</tr>
<tr>
<td>Eric</td>
<td>31-40</td>
<td>✓</td>
<td>5’6”</td>
</tr>
<tr>
<td>Elliot</td>
<td>31-40</td>
<td>✓</td>
<td>5’5”</td>
</tr>
<tr>
<td>Luke</td>
<td>31-40</td>
<td>✓</td>
<td>4’8”</td>
</tr>
<tr>
<td>Glenn</td>
<td>61-70</td>
<td>X</td>
<td>5’</td>
</tr>
<tr>
<td><strong>Women</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Bonnie</td>
<td>31-40</td>
<td>X</td>
<td>4’10”</td>
</tr>
<tr>
<td>Maddy</td>
<td>21-30</td>
<td>✓</td>
<td>5’3”</td>
</tr>
<tr>
<td>Judy</td>
<td>31-40</td>
<td>X</td>
<td>4’3”</td>
</tr>
<tr>
<td>Mina</td>
<td>21-30</td>
<td>✓</td>
<td>4’8”</td>
</tr>
<tr>
<td>Wendy</td>
<td>41-50</td>
<td>X</td>
<td>4’7”</td>
</tr>
<tr>
<td>Joanne</td>
<td>51-60</td>
<td>✓</td>
<td>4’9”</td>
</tr>
</tbody>
</table>
Figure 4 illustrates each theme and its associated sub-themes and I will use these themes to tell participants stories using their own words. Table 5 aids as a reference for additional participant information as age, height and treatment may give some context to participant responses. In this section I discuss how my research supports what is already known in the visible difference literature and highlight where my research is novel. I acknowledge that some of the issues discussed in this research are common to all and aim to tease out issues that are particular to people with RSS. This is a common challenge to researchers working in the field of appearance and visible difference (Rumsey and Harcourt, 2005a).

4.1 “It’s not just all about height”

The literature describes RSS as ‘heterogeneous’ (Penaherrera et al., 2010, Saal, 2005) meaning the clinical characteristics vary between individuals. In light of this I asked participants broad questions about how RSS affected them, any adjustments they made because of RSS and the affect it had on them over time. After asking these questions in the first few interviews it became apparent that the most homogenous clinical
characteristic, short stature was not the main concern for the majority of participants. I began to ask questions such as “If you could change one thing about your RSS what would that be?” and “What aspect of your RSS had the biggest impact on your life?” I also asked general questions about their experiences of school and college.

When answering questions about school and how RSS has affected them, participants answered in part by discussing their appearance. They used very negative words such as “horrific”, “unattractive”, “ugly” and “hate” to describe how they felt about the way they looked. Table 6 summarises the characteristics participants felt had the biggest impact and aspects of their appearance they found unattractive currently or in the past. I categorised the latter as ‘unprompted’ as I did not ask specific appearance related questions; these were responses that arose as part of a more general discussion about RSS.

Table 6. Characteristics of RSS that had the biggest impact on participants and unprompted appearance-related concerns.

<table>
<thead>
<tr>
<th>Participant</th>
<th>What has had the biggest impact</th>
</tr>
</thead>
<tbody>
<tr>
<td>Warner</td>
<td>Speech</td>
</tr>
<tr>
<td>Oli</td>
<td>Asymmetry</td>
</tr>
<tr>
<td>Wendy</td>
<td>Asymmetry</td>
</tr>
<tr>
<td>Bonnie</td>
<td>Asymmetry/scars on legs</td>
</tr>
<tr>
<td>Judy</td>
<td>Weight</td>
</tr>
<tr>
<td>Eric</td>
<td>Weight</td>
</tr>
<tr>
<td>Grant</td>
<td>Restricted leisure activities</td>
</tr>
<tr>
<td>Glenn</td>
<td>Height</td>
</tr>
<tr>
<td>Mina</td>
<td>Height</td>
</tr>
<tr>
<td>Joanne</td>
<td>Pain</td>
</tr>
<tr>
<td>Glenn</td>
<td>“Short and ugly”</td>
</tr>
<tr>
<td>Warner</td>
<td>“Wonky mouth” – asymmetrical face</td>
</tr>
<tr>
<td>Oli</td>
<td>“Unattractiveness of overweight” “bulbous” – asymmetry and weight</td>
</tr>
<tr>
<td>Mina</td>
<td>Small, but not in proportion</td>
</tr>
<tr>
<td>Wendy</td>
<td>“My teeth, that’s what’s held me back”</td>
</tr>
<tr>
<td>Joanne</td>
<td>“Angular face and hanging teeth”</td>
</tr>
<tr>
<td>Judy</td>
<td>“Physically unusual”</td>
</tr>
</tbody>
</table>
During discussions about school many participants described a time before they started school where they did not feel different at all: “I think certainly as a kid from 0 to 10? It was just who I was and that my horizon was very much kind of myself” (Todd). This then changed when they started school, began mixing with peers and started to realise they were maybe not quite the same as everyone else. Warner said “I’d be in the playground at school, would’ve been the first, probably the first time I realised that physically that I wasn’t as big”. Similarly, Glenn described how his appearance did not bother him until he was about 10 years old and it was at this age he began to realise the significance of his different appearance.

Glenn

*I didn’t like the way I looked. I didn’t actually look at myself in the mirror with the ability to analyse very quickly what I exactly looked like, until I was probably about 10 [...] and when I saw that, I didn’t like what I saw, because I was changing, and my puberty came quite early. And I didn’t like what I was seeing. And so I could understand why the girls liked me for my personality, but short and ugly, I don’t think so.*

The characteristic that caused the most distress was asymmetry\(^3\) (one side of the body is smaller than the other affecting the face and/or body) and was a common cause for appearance-related concerns. Participants’ issues with asymmetry ranged from every day, benign but problematic issues, to significant events that deeply affected their confidence and self-esteem. For example, Todd described the problems associated with having one foot bigger than the other and trying to buy and wear shoes “*[Y]ou sort of end up walking around feeling like you are some sort of pippy long stocking cartoon [a character that has odd stockings, one up, one down]*”. Bonnie described it as something that had bothered her in the past and that through a lengthy process she had managed to come to terms with how she looked. Bonnie was the only participant who had leg-lengthening surgery (although many talked about it being offered to them) and was also troubled by scarring on her legs as a result.

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\(^3\) A paediatric endocrinologist from the steering group for this research stated that he was shocked to discover participants had issues with asymmetry. It was not something he had even considered a possibility in his consultations with patients. This led me to consider the possibility that clinical consultations are not conducive for patients to discuss appearance related concerns.
Bonnie

“I think the asymmetry; it took me a long time, because I can really see it. And generally stuff with your arms by your legs and [...] the bad scarring on my legs; that’s took me quite a long time”.

Oli’s experience of living with an asymmetrical body had a profound effect on him affecting his ability to form romantic relationships. Oli recognised that this issue was not something others acknowledge as a potential problem “this is the whole thing about being the short person OK and also the asymmetry thing which people may not recognise but it’s true, it puts me at serious disadvantage”. Oli described a particular experience at a party where he was in a swimming costume and a girl at the party noticed his asymmetrical body and reacted negatively. Similarly, to Bonnie, Oli talked about his asymmetry as something he had to come to terms with, it was something that was beyond his and medicine’s control to change. This pivotal experience may have led to a short period of self-harming behaviour and lifelong depression.

Oli

Symmetry’s a good thing, it always has been. It’s the standard in everything else [...] that’s actually a big problem and people notice and that in my later teen years really got to me, a lot. To the point we were having some crazy party [...] and this one girl looked at me and she started freaking out because she was saying ‘what’s wrong with you, you look different, your left side’s bigger’ and then she just like freaked out because she’d just kind of noticed it for the first time and her reaction that I saw was very disconcerting [...] it’s not reconcilable, it’s not something you can get over or improve unless you somehow get the right side of your body bigger than your left side through some miracle of medical medicine[sic], other than that it’s there and there’s no cure and it has its effects.

Being overweight was another issue resulting in appearance-related concerns and for some it was their biggest concern. Weight is an issue common in the general population with 87% of women and 48% of men in their early 20s wanting to lose weight (Neighbors and Sobal, 2007). Being overweight was a more complex issue for these participants for
three reasons. Firstly, RSS patients are generally very slim; this resulted in some participants not feeling authentic in their representation of a person with RSS if they had gained weight. Secondly, some participants explained how it accentuated their asymmetry; for example:

Oli

A more horrific example would be when I started picking up the weight [laughs] and the left side of the fat on my body was like, bulbous was standing out more than the right side, I could see it and [it’s] this double whammy where there’s this thing about the unattractiveness of overweight and then this extra unattractiveness.

Thirdly, being short and overweight was not a winning combination “because when you’re short and being fat it makes you feel a lot fatter and shorter. If I was taller I could probably carry the weight a little bit, but I can’t” (Wendy). Participants constructed their own narratives as to why they gained weight. Judy felt that it was connected to her lack of mobility. Eric related gaining weight to his growth hormone injections as he could eat what he wanted during treatment but once he stopped he gained weight. Being overweight in adulthood had come as a surprise to some participants because as small babies they had difficulty feeding and their parents were encouraged to feed them high-calorie foods to help them achieve a healthy weight (Christoforidis et al., 2005). This has subsequently been found to contribute to an unhealthy BMI and other health related complications in adults (Ezzahir et al., 2005) and has been discouraged in the most recent recommendations for the treatment of RSS (Wakeling et al., 2016).

For Mina, height had a profound impact and affected her in a similar way to Oli. She was bullied because of her appearance, which may have contributed to her self-harming behaviour, chronic depression and social anxiety. Again, a combination of height and other factors seemed to cause additional dissatisfaction with her appearance.

Mina

I really hated the fact that I was little. I hated it in school, because I used to get bullied for it [...] It was mainly a height issue; I hated being small,
because I had really big hips as well, so I was just really short, with big hips and big thighs and I just...I hated being small.

Even simple things like having very small feet for Mina had repercussions “the nice pretty shoes that all the other girls wear [...] you have to go into the kids section and you’re like I don’t want trainers that flash up, because I’m twenty-five”. Being smaller than most people and having to wear children’s shoes or even shoes with the ‘back to school’ label led some participants to feel they were being infantilised.

For some it was not their height directly that had the biggest impact, but an artefact of being smaller than their peers. For example, Grant was not able to play rugby once his peers started to grow bigger. Grant was describing a common experience for male participants, whereby they were not only small in stature but also had a slim frame due to reduced muscle mass. Being small and slim was not conducive for these participants when playing contact sports such as rugby.

Grant

I guess the biggest impact was, well not the biggest, but one of the main impacts obviously I always enjoyed playing sports and from that point of view obviously used to play a lot of football, did play rugby at that time when I was still relatively similar to everyone else and then a lot of people just kept growing and I didn’t [laughs] so playing rugby came kind of out of the picture.

But for Warner his speech, resulting from a cleft palate, had the biggest impact on his life. He elaborated on the difficulties he had with his speech, he used the words “awkward”, “embarrassing” and “nightmare” when describing social interactions and how this aspect of his RSS has impacted him more as an adult “[W]hen you’re an adult you’ve got a job, you’ve got responsibilities and you’ve got to talk”. He also alluded to the possibility that different aspects of his RSS would have bothered him more at different stages “[T]he way I speak has more relevance than it would’ve done when I was younger”.

Warner
Russell-Silver is multi-faceted, it’s the main thing that people will notice will be the height, but actually the things that I notice are not the height, it’s other things. Yeah. That’s the one, if I could change one thing, it wouldn’t be my height it would be my speech [...] I’d say it has been a worse problem than my height.

Research on other short stature conditions support these finding that patients have concerns above and beyond height. In a study on women with Turner syndrome, Sutton et al. (2005) found that the most cited concern for participants was infertility with short stature coming second. Like other issues, height may be a bigger concern at different stages of the life span for people with RSS: depending on participant’s age, aspects that bother participants might change over time. However, in Sutton’s study they found that in a sample aged between 7 and 59, participant concerns remained stable across the life span. This may not be the case for participants in my sample, for example, Joanne found out she was infertile at a relatively young age which she described as “devastating” and now in her 50s it was her mobility and pain that superseded that impact.

4.1.1 Summary

“It’s not just all about height” illustrates that most participants had varied appearance and body image related concerns, above and beyond height, that led to them feeling visibly different from those around them. In contrast, participants’ experiences with health professionals centred on lack of growth and how to gain height. Participants’ descriptions of their regular hospital visits echoed this: “[I]t was very much about kind of taking blood and I remember a lot of my mum’s concerns and doctors concerns about growth charts and weight charts” (Todd). This particular discrepancy had not gone unnoticed by one participant who criticised health professionals and families for their continual focus on height and height alone. This participant stated that height is what people assumed a person would want to change whereas this is often not the case. One participant eloquently described that being short was not the worst thing that could befall a person. Once a child with RSS starts treatment to gain height they fall into the role of being a ‘patient’ who needs treatment to fix a perceived problem. But whose problem is it?
Judy

Being short is not the world’s worst thing. You know that, you have a child that’s short, but once you start all these interventions you then have a child that has issues and that is a sick child and needs treatments.

4.2 Resilience - threat and preservation

Participants described feeling different from others around them. While it is very common and not surprising to discover children with long-term conditions struggle with feeling different from their peers (Lambert and Keogh, 2015), health professionals working with patients or their families may not recognise that children and adults with RSS do feel different from those around them. Even though this is not a novel finding in the wider field of health psychology, it is a novel finding in regards to RSS research; therefore I am reporting these findings here. RSS is, for some, a long-term health condition and children and adolescents have regular hospital visits. Those that have growth hormone treatment may be on that regime from as young as two years old until they stop growing (16 years old for men and 14 years old for women (The MAGIC Foundation, 2016)).

I previously discussed how participants described what they perceived as undesirable aspects of their appearance. When they began mixing with their peers these ‘undesirable’ aspects became more apparent and, from the age of ten onwards, started to trouble them. Feeling different followed a similar pattern. When starting school ‘feeling different’ was neither good nor bad. This feeling remained until various external factors re-enforced their difference to the point they no longer felt untroubled. For some this came in the form of a statement of education, which meant they had extra help from staff or modifications such as a ramp instead of steps, as they were too small to climb them. It appeared that this special treatment was a constant reminder that they were not the same as their peers.

Warner

I remember during the first school, and probably through to middle school, there was certain things that were laid out for me that were different. [T]here was a ramp they built at my first school. I couldn’t walk up the steps, and I had
my own dinner-lady specifically designed to look after me. So things like that I guess would have made me feel that I was a bit different.

As participants moved into adolescence the differences they felt began to trouble them: “it’s a realisation that there’s something wrong, that whole thing that you’re different is actually not as you think” (Oli). Participants’ confidence and self-esteem were affected by feeling different. Warner described feeling “inferior” to others and that being treated differently at school became “self-fulfilling”: because he was treated differently this reinforced and validated his thoughts that he must be different. Grant also felt that feeling different affected his confidence.

Grant

I mean socially I think I was never when I was younger I didn’t have much confidence and maybe that was a knock-on effect from that knowing and feeling maybe different and I was always quite shy growing up till a certain age.

Also around this time some participants started their puberty early, which does sometimes occur in people with RSS (Wakeling, 2011). Men viewed this as a positive change; “I reached puberty earlier than my peers about a year or so, being a boy that wasn’t such a bad thing for me [laughs] at the time it was kind of cool or whatever” (Oli), but for women this change was unwelcome. Maddy described starting her periods earlier than her friends as “traumatic” and said that “if no-one else is sort of developed and things, you become more self-conscious about it”.

Feeling different was not an experience exclusive to adolescence, as Warner explained: “I guess a lot of people see me as a bit of a novelty character”. When socialising with friends for example, people would ask to have their photo taken with him. Judy felt more different as an adult compared with when she was a child: “There was times you know I did think sort of why am I different, and I think it’s been more as I’ve got older”.

Feeling different led participants to try and fit in with society and with others with RSS. I asked Grant if he felt wanting to fit in was related to him having RSS and he said “Yeah
probably actually yeah wanting to fit in [to] I hate the word normal, there’s no such thing, but you know try and be similar to the other kids and not stick out as being too different”. Participants talked about wanting to be ‘normal’ but found it hard to define what this was.

Participants expressed that having RSS and feeling different from those around them led to a range of experiences, which varied from finding disclosure a challenge and not telling anyone at all, to being proud and open. Some participants kept it from their friends, for example Elliot called it a “secret” that he was “fearful would become a thing”. He felt that it might define him if he told friends and work colleagues. By not disclosing he could retain a sense of normalcy, though non-disclosure may put resilience at risk as supportive relationships are key here (Samuels and Pryce, 2008). Bonnie said she was in denial about having RSS and tried not to acknowledge its existence until the symptoms got unmanageable. Maybe by not acknowledging it she thought it would never affect her “[A]nd my health just got worse and then that’s when I thought I could do with knowing more about it because I think I’d just hidden away from it all my life” (Bonnie). For some, not disclosing appeared to work well for them. But for others, keeping RSS hidden and the impact their condition had on them, took its toll psychologically. That, and the shame of having RSS could have resulted in mental health issues that made it hard for them to talk and seek help.

Oli

But that when all of this came up [...] the shortness, the asymmetry, it floored me, I couldn’t deal with it. [S]o yeah and the worst thing in hindsight was that I didn’t tell anyone.

Some participants were proud of having RSS and of the person they had become because of it. Todd described a conversation with his girlfriend about the possibility of having a baby with RSS, which was not an issue for him; he felt that he had to defend himself and his syndrome. He would not change who he was, had never considered finding out if there was a risk of passing on this genetic condition to his children, and would have been happy to have a child with RSS. From what Tod described it appeared that even though he
had experienced adversity, he was happy with his life and who he was, and felt that having a genetic condition had contributed to that.

Todd

*And I remember getting quite defensive about it. You know, being like, well, I don’t, it’s never really affected my life in a negative way it’s an important part of what I am and I kind of be quite proud to have a little child that had RSS.*

Others did not share this positive view. Oli used words such as “ashamed” and “disappointed” when describing how he felt about living with RSS and said “Ah dam. I got this card”. Others started their interviews by saying that they were not affected by their RSS, but as the interview progressed they revealed times of adversity relating to their condition and described times when they felt visibly different. Reasons for this could include: dismissing the extent to which their lives had been affected by RSS as a way of coping, or a way of normalising the impact RSS had to preserve self-identity and self-esteem: “Well like I say it’s never been a big part of my life. It’s not even been a little part of my life really. You know I was born with it yeah, and I’ve just got on with it.” (Wendy).

Feeling different also led some to access support. Participants discussed using support groups for reasons such as: knowing they were not alone, finding out others had similar symptoms and experiences to them, and that they were not a unique “mutation” (Judy). Some had always been part of the RSS community as their parents had accessed this support and attended yearly conferences, whilst others made the decision in adulthood to seek out support as their symptoms had changed or they had become curious about other’s experiences. Some described being nervous about meeting others with RSS as they had never met anyone else similar to them, but after joining the group they felt comfortable there. Others felt that they could not or should not access the support network available because, in their opinion, they did not have severe enough symptoms and would be judged by others in the group.

Maddy
Joining some of the groups I’d feel slightly like a bit of fraud on it, because I’m nothing like really what some of the other people who’ve got it quite severely. I’m quite on a different kind of spectrum of it I think.

Some participants reported finding it hard to discover similarities between themselves and others with RSS, which left them in an indeterminate state with no one group to belong to. Some felt their RSS was not as ‘severe’ so consequently felt like an imposter in online support groups. One participant described not fitting the typical RSS ‘look’, which made her feel that parents of children with RSS would judge her more harshly when she met them at conferences than the strangers in the street.

Judy

[S]he’s overweight but our child’s really skinny and I thought Russell Silver adults were skinny [...] I think sometimes the actual parents here can be more judgemental actually thinking about it than people outside, because to other people outside I’m just short and overweight, but here they’re thinking ‘My child could be like that’ or ‘I didn’t think Russell Silver adults got overweight’.

Accessing support groups appeared to facilitate downward social comparison as is demonstrated in Maddy’s quote above, which may have had psychological benefits (Festinger, 1954) and enhanced self-esteem and resilience (Taylor, 1983). Todd stated that comparing yourself to others is something that everyone does and that although he could see he was different, he felt neutral about what that meant: “you inevitably begin to compare yourself a bit more to other people (pause) and sort of I saw myself a bit as different but not necessarily any better or worse”. A common thread for downward comparison was that it could have been much worse. However, for some participants it was not as straightforward, for example, Oli talked about feeling glad he was not as affected as others whilst also acknowledging he was more similar than he wanted to admit.

Oli
I’m not sure if I felt lucky I wasn’t one of them or unlucky that I was one of them. Sort of a mixture of the two [...] I sit and think that I’m almost something between what people would call normality and this group and for me that’s even more frustrating because I can’t I can’t identify myself with them, but I can identify with them.

Accessing online support also seemed to provide opportunities for downward comparison. Wendy used a Facebook group that enabled her to see she was “one of the tall ones”. She seemed to have contempt for those that appeared needy and reliant upon one another and did not identify herself as someone who needed support. Also some participants had accessed online academic journals which allowed them to see which characteristics they had compared with participants in the report and to see it could have been “much more dramatic” (Elliot) for them.

Whilst downward comparisons were often made to others with RSS, upward comparisons, which may be associated with negative psychological consequences (Festinger, 1954), were more commonly made with those without RSS. Often comparisons were made to siblings who were described as either more successful at sports, in their careers, or possessing more physically attractive qualities. Participants described dealing with this either by losing interest and taking up a different activity that required less physical effort, taking some comfort in characteristics such as intelligence that enabled them to feel on an equal footing with peers and siblings or by trying harder and competing with those around them.

Oli

I do think one of the things that did impede is for me to do sports ‘cause my brother is a world first team rugby player my sister has performed and did all these things so I don’t wanna be self-deprecating and say I was the runt of the family because that’s probably not the right word but in a way there’s a lot of things that I couldn’t do or didn’t want to try or wouldn’t be the best at that [...] it did make me unhappy, discontent in a way. I think the way I compensated for it was by just losing interest, so because if it’s not important then it doesn’t matter.
identified in the data that participants were using characteristics other than appearance to compensate for looking or feeling different and to protect them from the judgement of others. They made an effort to build friendships, using their humour and intelligence to build confidence and self-esteem and to garner social capital. Many participants talked about their visible difference not stopping them from making friends; “[O]ne thing that I think Russell Silver has never held me back on is my social life” (Warner), that they had a large social group; “I always had lots of friends to play with” (Glenn) and that they consciously invested time and effort in building friendships as a way of fielding negative consequences: “[I] probably tried too hard sometimes to fit in too much or you know please everyone or wanna be everyone’s friend” (Grant). Others described using strategies, such as “making people laugh” (Glenn) as a way of gaining friends or as a buffer against adversity: “I had to learn to use my mind pretty quickly and a sort of sense of humour as a way to kind of survive” (Todd). Humour may play an important role in resilience and psychological well-being and there are thought to be adaptive (self-enhancing) and maladaptive (self-defeating) styles of humour (Kuiper, 2012). Though participants appeared to use humour more as a social lubricant to develop and maintain social connections. Others focused on their strengths, such as intelligence, as a way of building self-worth and making areas they felt self-conscious of “less relevant” (Oli).

There may have been a down side to some friendships and social interactions. Whilst some described befriending older or bigger children as protection against bullying, others felt more comfortable with children younger than themselves. And one participant described being asked by strangers if they can have a photograph taken with him on nights out or being lifted up frequently. Balen et al. (2006) warned that becoming a ‘mascot’ or socialising with children younger than them may be a result of negative self-judgement rather than being seen as sociability.

As well as humour and intelligence, participants used sport to overcome feelings of inadequacy. Some participants struggled as sport made them feel less able, but some used sport as a way to “find some parity with lots of other big kids” (Todd). Participants described overcompensating, for example being overly aggressive when playing sport, to make up for their size, being overly assertive: “I felt that I had to exert myself on
situations” (Eric) or having accessories that indicate higher status, such as an expensive sports car: “I wanted to show people that there was more to me than what you saw” (Luke).

For those with visible differences environmental factors such as comments from peers and strangers can have, for some, a significant impact on self-esteem (Williamson, 2014). I specifically asked participants about their experiences of bullying and stigmatisation. Fortunately, it was an uncommon experience among participants and only Joanne and Mina explicitly described incidences of bullying. Mina was bullied about her height and found it hard to tell people why she was small as she felt they would not believe her or understand. Joanne was bullied constantly in late adolescence because of her appearance.

Joanne
I think as a child obviously bullied a lot, because you’re in that transitional stage of looking different, your face is angular and different [...] and you’re a lot smaller than all the others. When you’re at Primary School you kind of, I think you kind of look a bit cute, it’ll be okay; as you get older you don’t [...] secondary school was the hardest I think for me [...] but it was definitely the early teens that was really, really bad [...] But I think Secondary was where it was so constant you couldn’t, so many, so constant you couldn’t avoid it.

More commonly participants described times in adulthood where they have been subject to unsolicited questions, comments and staring. Participant reported being stared at when they went out and hearing comments such as “Oh what horse are you riding, I’ll be backing it” (Warner) or “Mummy, mummy why is that lady short?” (Judy). Participants did not seem overly affected by comments and staring, understanding that it was natural for people to stare at anyone who looked different. They chose to interpret other’s motivations as curious rather than malicious: “I’ve always tried to - if anyone laughed at me or anything - I try to show them the human being within me and be helpful towards them” (Luke).
Employment came up as a difficulty here with participants perceiving employers having lower expectations of them, which had an impact on career prospects: “the career thing I think in my case (pause) I’m pretty sure that it does affect how people turn to you and think of you” (Oli). Luke turned this to his advantage: “the lower they are [expectations] the more scope you have to impress and come out looking better than everyone else around you. You learn to work that ignorance and that naivety to your advantage and then you come out on top” (Luke).

When answering questions about their experience of healthcare, most participants reported that they felt well cared for, but there was a negative side to their regular hospital visits. Wendy described feeling “humiliated” by having photographs taken in just her underwear, one participant described how having his testicles measured led him to wonder if something was wrong or that he was different to others, another stayed in hospital for weeks at a time and suffered abuse as a result.

Glenn

I went back to the hospital [...] and that’s when things went really nasty [...] the Sister who ran that ward [...] was extremely strict [...] she was quite violent, verbally [...] it was when the needle went in that it really hurt, and you screamed and [the Sister] would always be somewhere around the bed telling you to shut up. It’s a long time ago. The NHS wasn’t very old.

Luke’s memories of a visit to hospital were worrying and he lived with the repercussions of this experience for many years. He was told by his paediatrician that if he did not have growth hormone therapy: “I would probably not find a relationship and that I would find it very difficult”. He felt as though this comment contributed to his lack of confidence in finding a romantic relationship: “about 3 years ago I had CBT therapy and basically I realised through that that what I’d been told about relationships what I’d been told growing up, reminds you of the fact that at 38 I hadn’t had a relationship and it all came from that comment probably at 15”.

Experiences of health care were not contained to adolescence. Participants had two main issues with the healthcare they received in adulthood. First, they felt neglected once
becoming an adult, as regular check-ups and hospital visits stopped abruptly: “once you leave your childhood [and] become a normal citizen, the NHS moves on” (Oli). For one participant, the constant monitoring during childhood and adolescence, led to health anxieties during adulthood. He described catastrophising a headache into a brain tumour because his health was no longer being monitored; he worried that he could become ill and no one would be there to identify it. And second, many reported going to their GP with symptoms that would always be labelled as RSS: “If they’re presented with something and they don’t know the answer, they’ll just say that’s just due to Russell Silver Syndrome” (Bonnie). This was significant because participants were worried that if their symptoms were not related to RSS there could be an underlying cause that was not being investigated. Both these issues led participants to request an adult point of contact, someone who could validate their experiences and relieve their anxieties: “just sitting in front of somebody and they know all about Russell Silver Syndrome, to me that’s priceless” (Judy). Often participants found non-specialist health professionals did not take them seriously.

Wendy

I think that for me is the most difficult bit, because you haven’t got anywhere you can go and say tell me what’s wrong [...] You know I think with the GP, so it’s not really discrimination, but it’s a knowledge of not knowing [...] So you kind of feel that the doctors are getting a bit cross at you. And by the time I go for tests, the inflammation has all gone down, so nothing ever shows. Because it kind of makes you feel a bit like a hypochondriac [...] he told me to get dressed, stop wasting his time and go home because he’s got patients outside that are in more pain than I am.

Adversity such as feeling different, comparing themselves unfavourably to those around them, feeling as those others were judging them based on their appearance, and having a genetic condition, affected some more negatively than others. Participants candidly shared times in their lives when they had experienced depression, anxiety (panic attacks, social anxiety), which for some led to self-harming behaviours. Participants directly related the way they felt about their appearance to their psychological distress.
Mina

I went through a very low time and I was self-harming, because I physically hated the way that I looked, I hated myself and knew there was nothing that I could do about it to change it, and I couldn’t accept the fact that was who I am.

Their distress seemed to centre on others judging who they were and what they looked like. Some participants did not realise it at the time, but in retrospect they could see that what they thought was sickness or low blood sugar was, in fact, what they believe now to be anxiety and panic attacks. Much of participants’ psychological distress began in secondary school around the ages of eleven to sixteen, with the first episode of depression occurring in their late teens. This pervaded into their early twenties and for some into their thirties and forties.

Luke

[I]n my teens I used to have a mini breakdown about every three months and I would cry. I think it’s quite interesting, it was literally about every three months where things just got on top of me and I didn’t know why, what the emotion was or why I was crying or anything, I just felt why am I like this [...] I think I felt sorry for myself [...] Not having a relationship. I didn’t have many later on in life.

Participants attributed this distress to a variety of reasons relating to RSS. For example, having negative feelings about the way they looked and who they were or being told certain options were not open to them because of their height. Wendy’s careers advisor told her that because she was in a low maths set she could not work in a shop and “I couldn’t work in a factory because the teacher said I wasn’t tall enough to reach a conveyor belt” so the only option was office work. She said “I worked in an office, which I absolutely loathed, and I think that was when I became really nervous. And on my first day at work I had a panic attack and ended up in hospital”.

Participants gave examples of times when they were socially anxious and would avoid contact with friends as a way of managing fear of what others may think. Due to
gastrointestinal problems relating to RSS, one participant would often be sick in the night as a child. Too embarrassed to have friends over to stay or to stay at theirs she described colluding with her parents to tell friends she was grounded as an excuse. Another distanced himself from friends and family: he did not confide in anyone about his RSS or his psychological distress and pretended that he was fine to also ensure people around him would not ask questions. He acknowledged that people may have noticed, but he ensured that a distance remained between him and others.

Oli

I kept it inside. I can hide it quite well. I’m good at that. I’ve been well trained I guess. I’m sure they noticed, they just didn’t ask. Again I kept myself from people as well [...] I didn’t speak to anybody and I didn’t interact with anybody else.

I identified points in participants’ lives where adversity would peak or they would experience acute periods of distress triggered by certain events. Again, often difficulties occurred during adolescence, which was also the time when some found out they were unable to have children of their own. Judy had chronic depression, and found out she was unable to have intercourse without a surgical intervention and having children would have to be through surrogacy. I asked her how she coped at the time with that news and she replied “Throw myself off the nearest bridge was the first [thought]”.

Feeling different was not always negative, for example, Maddy talked about feeling “special” when she was younger and that she got attention even though she was not ill: “I felt a bit special I think, which is a bit odd, but that’s sort of because there was nothing actually wrong with me, physically, but occasionally I got a bit of special extra treatment when we went on trips to London”. Also, participants tried to positively reframe RSS and the difficulties it caused them. Judy was born without a womb, but still managed to see the advantage in this: “And from hearing about women having problems with their periods and all the rest of it, I don’t have any of that. So on the positive you don’t have to worry about pads and embarrassing problems and your time of the month”. She also talked about being shorter than many others with RSS as an advantage. Because they are closer to average height, she reported, others find it harder to believe they have a health
condition, whereas for her it is obvious: “I can say I’ve got problems and it’s part of my [RSS], then it’s accepted, so I think some have a harder problem”. Is it possible that those less effected by RSS face a unique challenge as it is easier to be who you genuinely are if your disability is more obvious to others (Waldboth et al., 2016).

4.2.1 Summary
Fletcher and Sarkar (2013) reviewed the many definitions of resilience in the literature and concluded that they all centre on two main concepts: adversity and positive adaption to that adversity. Participants described adversities such as feeling different to others leading them to compare themselves to those around them; and in turn, others judged them on their appearance and abilities. Comparisons began to be drawn in early adolescence and contributed to issues that extended into adulthood. Young people are more likely to make comparisons during this time as they are going through an important developmental phase during adolescence and spending more time with their peers (Myers and Crowther, 2009). Due to their appearance-related concerns these participants may have been more likely to compare themselves to others as research shows a strong relationship between body dissatisfaction and social comparison (Myers and Crowther, 2009). Other adversities such as early puberty, feeling shameful, staring from strangers, and difficulties dating and maintaining romantic relationships, were also discussed. Many strategies were used to cope with adversity, for example, using support groups, social comparison and fostering a positive identity. They may have used strategies such as non-disclosure, social avoidance and capitalising on other characteristics to avoid being bullied and/or stigmatised. Some of these strategies could be described as adaptive and some maladaptive, the former leading to resilience and the latter to psychological distress.

4.3 ‘Mayor of the friend zone’
I asked all participants to tell me about their experience of romantic relationships because I had asked about other kinds of relationships (friendships/siblings/parents) and I struggled when deciding how to label the type of relationship in this theme; was it romantic, sexual, intimate? I settled on the term romantic relationship, but acknowledge that others may find it unsatisfactory. The reasons I chose ‘romantic’ were: it was a common term used in research with adolescence, romantic relationships and visible differences (Giordano et al., 2006; Ambwani and Strauss, 2007; Bentley et al., 2007; Sanchez et al., 2008; Collins et al., 2009; Griffiths et al., 2011; Smiler and Heasley, 2016). And secondly, ‘intimacy’ has been used in research to describe aspects of a romantic relationship (Bell et al., 2015; Sharratt, 2015) and thirdly, as Stock et al. (2015) found with their participants, they were striving for a romantic relationship rather than just brief sexual encounters due to placing more value on personality and companionship rather than just appearance.
not because I had any evidence that there would be issues. I did find there were issues for male participants particularly in forming romantic relationships and as I did not anticipate these answers I did not delve deeper into these responses and ask questions about intimacy in romantic relationships. This lack of perceived or enacted romantic appeal led to participants to feel as though they were “mayor of the friend zone” (Todd). Fogg (2013) wrote about the ‘friend zone’, which he defined as ‘an ostensibly platonic relationship in which one person is romantically interested in the other’. This theme is exemplified by the following quote:

Todd

I think one area where it did really did have an impact was with girls in that teenage phase [...] I think I had pretty low sexual self-esteem all the way through my teenage years and actually probably through most of my 20s [...] I was very definitely mayor of the friend zone, I’d become very good friends with all of these very beautiful girls [...] I really struggled with forming relationships.

Todd described how his issues with appearance impacted significantly in adolescence. Not being able to form romantic relationships started in adolescence but also pervaded through his twenties. Due to low confidence and self-esteem he almost put himself in the friend zone. This issue was common among men, but not exclusively, and problems for some continued into their late 30s. Some participants did not put themselves in the friend zone but were placed there by trying to become more than friends and then feeling rejected when this was not reciprocated “I suddenly realised I liked them (girls), but they didn’t seem to like me when I wanted to like them a bit more [...] I had lots of girlfriends, but no girlfriend [...] I felt rejected [...] but they used to love talking to me” (Glenn).

There were many examples in the data where men appeared to feel that they had fallen short of the masculine ideal and in particular what a male body looks like. A common issue was with muscularity; participants felt that they were not as muscular as other men and this is a common characteristic of Russell-Silver syndrome. Research has shown that many men are unhappy with how muscular they are. Tiggemann et al. (2007) found that
in a sample of 119 heterosexual men aged 18-60, when asked about their ideal body, 80% wanted to be bigger or more muscular and that ‘[T]he male body ideal involves being thin and being muscular’ (Tiggemann et al., 2007, pp. 22). This issue is heightened in men with RSS as; because of their genetic condition they find it very hard to become muscular.

Warner

*Because of my height, I guess most girls are looking for guys who’re a bit taller. And my lack of muscle. Girls are looking for guys that have sort of got a bit more muscle. Not every girl. But generally if you take a hundred random women, how many of them, if you stick in a guy [...] six feet tall with average build, and you take me, you’d be lucky if one of them picks me over the other [...] and that’s just reality.*

Warner described the combination of being slim and short as not the type of guy girls want to date. Participants described other issues with body image and appearance that may also be related to how masculine they feel and how others perceive them. Todd said that RSS was “*slowing me down from growing up*”. He was in a hurry to become an adult and as he saw it becoming a man involved being big and hairy, but he was “*small and skinny*” and looked five years younger than his age.

Todd

*Then I think sort of through that the late twenties phase with the relationships I think I probably saw myself as being worse, as being sub-standard, being less of man in a traditional sense and also kind of being in an environment where the idea of what a man was, was quite narrowly defined as being kind of you know big and muscular and sort [of] hairy.*

The data revealed that many male participants did not perceive themselves to have ‘masculine capital’ and perceived that others felt the same. Masculine capital can be accumulated and lost through certain behaviours and assets that build up gender identity (De Visser et al., 2009). Participants may have tried to transfer masculine capital from one area to another, for example, their perceived lack of masculine physical attributes may have resulted in over-competitiveness, participation in sports, and taking the lead in a
dating situation. Masculine capital in men with RSS may not just hold them back in relationships but also in other areas of life. One participant speculated that other men he worked with may have been more successful than him because of masculine attributes they possess that give them ‘presence’, such as a deep voice and height. He did go on to discuss that it is more complicated than that and he finds this path of thinking to be superficial, but nevertheless it was a conclusion his mind jumped to when he was feeling vulnerable about himself.

Elliot

_ I know that there are, even today, when I go back to the office there are people who have been significantly more successful than I consider myself to have been, and I look at them and the first thing that comes to mind is well hey that’s a tall bloke and he’s got a deep voice and he has great presence about him._

Other participant’ masculinity may have been affected by the reaction of family members when they were children. For example, Glenn described feeling that he was never the son his father had hoped for. His father was called home from working away because he was a sick child with RSS, so having a medical condition and being small for his age made this participant feel as though he was a disappointment: “_My father, I think he felt very disappointed to have a child that was so small and feeble, in his eyes weak._”

Participants discussed other, more behavioural, elements that appeared to be attached to their sense of masculinity or could enhance their masculine capital. Sport was a topic that came up often; some participants were unable to participate due to their size and strength. But those that were able to participate saw it as an opportunity to show that they were the same as everyone else; to gain some masculine and social capital: “_the big turning point for me was actually getting stuck in with some of the football team, it almost felt like you were accepted_” (Grant). Playing sport lead to a feeling of equality among other men, but there was also a perception that playing sport is a desirable characteristic for romantic relationships: “_Even at that age I feel like the football players or the taller blokes or stronger perhaps did better with girls than those who were perhaps shorter or fatter_” (Elliot).
Participants were very candid in what they discussed. One participant explained that RSS might have affected his sexual performance in the past. It is possible that other participants had similar experiences but did not feel they could discuss it and I did not ask the questions specifically.

Fogg (2013) purported that men who get stuck in the friend zone are probably those with low confidence and self-esteem, they are too shy to make a move. He made the point that gender roles may exacerbate this issue further as generally men are expected to make the first move. This was the way Luke gained masculine capital back for himself when starting to date in his late 30s: “‘Is it ok to do this?’ no say ‘I thought we could do this. Is that ok with you?’ You know what I mean, I was taking the lead, I was having the role of a man, knowing what the role of a man [was]”. He decided how a man should act around a potential romantic partner and emulated that.
“Mayor of the friend zone” was a more common experience for men, though a few women reported similar experiences. Wendy’s quote below echoes that of Todd’s and describes how she really wanted a boyfriend, but they saw her more as one of them.

Wendy

I was quite boy mad actually you know, when I think back. I just didn’t have a lot of boyfriends. They all liked me, but just not that way. I was a bit of a clown really I suppose. I was one of the lads.

Wendy did have one relationship during secondary school, but had to keep it secret because “he was worried about what everybody else was going to say”. She said that he only went out with really pretty girls, but liked her. This must have been a bitter-sweet relationship as she really liked him and was flattered he wanted to go out with her, but he was too embarrassed to be seen by others with her.

There was one female participant, Judy, who had never had a romantic relationship. Similar to others, she had male friends at school but no boyfriend: “I had sort of male friends that I was close friends with”. However, the circumstances that lead to this were different in part than those experienced by Wendy and the other male participants I have described above. Judy had gynaecological issues, which were discovered in adolescence after she did not start menstruating “they found I had no opening to my vagina”. She described the issues around romantic relationships and disclosing this information to a potential partner.

Judy

I think it’s difficult for me because when do you tell somebody? You know you don’t tell them on the first date but you don’t tell them before the wedding, you know you don’t leave it until the last minute either. But you don’t want to scare them off or you don’t want to discuss such intimate things straightaway. So you know and I don’t scream it from the rooftops that I have these issues.
Stature for men may be bound with other attributes that contribute to masculine ideology, but being small and female may be an advantage to the sexual side of romantic relationships, as one female participant describe it “[G]uys love it when you’re small. They do, because they’re just like ‘Yeah, I can just throw you around’” (Mina). When Maddy was asked if RSS affected her romantic relationships at all she said, “No, I don’t think so [...] if anything it’s, you know you’re littler, so you’re seen as kind of a bit cute and it’s a bit quirky and that sort of thing”. Maddy described an earlier time when - because she was on GH therapy - she was bigger than her peers. Although Maddy's experience is unique among this sample, it illustrates the difference between men and women and how stature affects them.

Maddy

It’s not very feminine I suppose to be the bigger girl and the taller girl, and I think I developed much earlier than all my friends did. So I was quite, self-conscious I think in that sort of stage, and I think that’s impacted my confidence and things.

As with the idea of masculine capital, women in the study did describe aspects of their RSS that led to them not feeling as feminine, take Maddy’s quote above. She also expanded on this point by saying: “they’re sort of sweet and petite, and you’re sort of not”. Comments made about their appearance also indicated some women felt they fell short of the feminine ideal “[Y]ou know because I’m not the tall leggy blonde with blue eyes that you know a lot of men would want” (Judy) and “I didn’t have big boobs and blonde hair and big eyes and you know that really stand out” (Wendy).

Another aspect of RSS that may affect participants’ feelings of femininity is infertility. Several women in the study could not have children: “No, I mean tried for kids, it never happened” (Joanne). When asked how it was at the time to be told that you cannot have children she said: “Well the best word is devastating”. Judy also described that time in her life as one of “the hardest times that I’ve had”. For Mina it was another aspect of her RSS that led to psychological distress, being told she may not be able to have a child was devastating as being a mother was her main aspiration.
Mina

*When I was in school, they were like what do you want to be when you’re older, I was like I want to be a mum. That’s all I’ve ever wanted to be, and it got to the point I got so depressed about it, I couldn’t watch babies on TV, I’d have to turn the adverts over, I got so down about it, it was unbelievable.*

Both men and women may have had high levels of rejection sensitivity. If a participant was unhappy with their appearance either due to their own perceptions or the perceptions of others “*Why would anybody like this, why would anybody want to go out with this!*” (Mina), they put themselves in a situation where they could be rejected, they resigned themselves to the fact it was because of the way they looked “*You completely understand why you are being rejected and you would have rejected them you know I’d have rejected me if I was myself!*” (Todd). Participants conveyed this idea that they could not have relationships with people they liked because in a sense they felt they were “*sub-standard!*” (Todd): “*When I already know that if I was that person and I had standards I would be automatically excluded!*” (Oli). But this did not stop them from having standards themselves and being unsatisfied with second best “*So it would take somebody with a special heart anyway to be with somebody who is physically unusual!*” (Judy). That person would have to be extra special to compromise their standards to be with someone who looks different. These beliefs are similar to others with appearance-related concerns as these beliefs affected communication with potential romantic partners, confidence in initiating contact and fear of rejection (Griffiths et al., 2012).

The data showed that not only had participants experienced challenges with forming romantic relationships, but also once they had found love it may not have been as they had hoped. One participant described not being happy with her first husband and that they did not have a sexual side to their relationship. She had continued, despite wanting to end the relationship, to please her parents.

Wendy

*Obviously with me growing up they maybe thought I was never going to find anyone [...] because then I said I was going to get married my mother ran*
away with the idea and she was then arranging everything and paying for everything and then it got so I daren’t say I didn’t want to.

Another had resigned themselves to the fact that he could not have what he wanted “I always had this idea of a perfect relationship and I can’t have it because I’m not perfect” (Oli). And another described that he is in a casual relationship that he is happy with but appeared to prefer to be in a more stable or committed relationship: “I think my ideal scenario would actually be to find someone in later life maybe. Maybe that’s what will happen” (Warner). These participants are describing at times settling for second best. If your needs are not being met in a social exchange this may have a negative impact on confidence, self-esteem and overall satisfaction.

4.3.1 Summary
This theme encapsulates the experience of many male participants – and a few female participants - in this study who did not perceive themselves to have feminine or masculine capital and perceived society to think the same. Masculine capital could be seen as similar to social capital and describes the resources a person has related to what is culturally or socially determined as masculine (Balen et al., 2006). Low masculine capital may have created difficulties in forming romantic relationships in particular; other social relationships, such as friendships, seemed to flourish for these participants. Friendships were the ‘silver lining’ to this theme as the majority of participants had close and supportive friendships that may have led to strong foundations to build romantic relationships in the future. This theme also identified that looking different did not just affect participants’ perceptions of themselves and their chances of finding satisfying romantic relationships, but health professionals and family members shared the same concerns.
5.0 Discussion

The aim of this research was to explore what it is like to live with RSS. I interviewed 15 adults and analysed the data using thematic analysis. Three themes were identified: “It’s not just all about height”, ‘Resilience – threat and preservation’, and “mayor of the friend zone”. This study is the first of its kind to explore how RSS affects people in adulthood and to conceptualise what it is like to live with this syndrome. This study has shown that people with RSS can experience fulfilling and successful lives. Participants expressed that they were all generally happy, had successful careers, were working towards achieving their goals, and had families, or were starting to have families of their own. However, there were significant challenges of living with RSS in adulthood and many of these stemmed from challenges in adolescence. Feeling visibly different from others, being unhappy with their appearance in adolescence, and others focus on gaining height, resulted in psychological distress and difficulties forming romantic relationships, especially for men. These difficulties pervaded into adulthood as some participants had still not managed to forge and maintain satisfactory romantic relationships. What this study also shows is that, contrary to previous literature suggesting that RSS has a negligible impact in adulthood, people with RSS experience psychological and physical issues across the life span.

The findings from this study mirrored many aspects of other conditions, such as Achondroplasia and Turner syndrome. However, my study identified unique challenges, such as appearance-related concerns in respect to the binary concepts of femininity and masculinity. My findings parallel research on visible difference in more respects than research on chronic illness and disability, which supports previous research showing that people with short stature identify more with being ‘different’ than ‘disabled’ (Shakespeare et al., 2010). The experiences of people with idiopathic short stature, Achondroplasia, Turner syndrome and Constitutional Growth Delay also echo less with the literature on chronic illness and more with that of visible differences. These findings contribute to areas of health psychology that are currently under-researched, such as, how visibly difference impacts romantic relationships and appearance-related concerns in men.
In this section I present key points from the findings for discussion and how these relate to previous literature, make recommendations for practice, discuss the strengths and limitations of this study and outline future directions for research.

5.1 Key discussion points from findings

5.1.1 Adolescence was a difficult time
Challenges during adolescence were central to all three themes and is a time when physical performance and appearance are important (Claessens et al., 2005). Although the challenges described below began in adolescence, for some, these challenges pervaded into adulthood. Participants reported feeling different from their peers during this time. By not disclosing RSS to others and not acknowledging they had a long-term health condition, participants were trying to live life in the same way as everyone around them. Often children will not disclose health conditions to maintain a normal life to outsiders and also to themselves (Kirk, 2010), though by not disclosing their health condition to others friends will not know they need extra support. Capitalising on available social support could reduce the impact of stress and aid coping in children with short stature (Balen et al., 2006) and having the involvement of friends can support children’s adaption to the challenges living with a chronic illness brings (Greca et al., 2002).

Participants compared themselves to others during adolescence, which led to strategies such as employing humour, intelligence or athletic ability to compensate for areas in which participants felt they were lacking. Researchers have identified this as a strategy used in other conditions that effect appearance (Balen et al., 2006, Stock et al., 2015, Williamson, 2014). This strategy may be adaptive and lead to positive adjustment as those that value attributes, other than appearance, experience less psychological distress (Thompson, 2012, Williamson, 2014).

If bullying did occur, it was more likely to happen during adolescence, supporting the findings from previous visible difference studies (Almenara and Jezek, 2015, Buhlmann et al., 2011, Griffiths et al., 2012, Lovegrove and Rumsey, 2005, Magin, 2008). Most participants may not have identified with being bullied, but had been subjected to
comments, staring, discrimination and stigmatisation. And although most participants did not report being unduly affected by comments and staring, they did report feelings of shame, secretive behaviour, self-harming, depression and anxiety. This may indicate that they were more affected than they wished to acknowledge.

During adolescence many male participants, and some female, reported difficulties forming romantic relationships. Similar results were found in Turner syndrome (Boman et al., 1998, Carel et al., 2006, Kagan-Krieger, 1998, Sutton et al., 2005) and Achondroplasia (Gollust et al., 2003). There are several possible explanations why participants experienced difficulties forming romantic relationships. Firstly, particular characteristics of RSS (short stature, low muscle mass, infertility) together with more general appearance-related concerns may have resulted in a perceived lack of feminine or masculine capital. Feeling as though they had fallen short of the typical feminine or masculine ideal may have eroded participants’ confidence and self-esteem. Men being more affected by appearance-related concerns was also found by Ambwani and Strauss (2007). They used qualitative methods to investigate the relationship between body esteem and romantic love in males and females and found that males more often felt that negative body image had made them hesitant to pursue romantic relationships.

Participants may not have been as successful at attracting a romantic partner because of their asymmetry. A preference for symmetrical faces may be related to sexual selection and mate choice in males and females (Griffey and Little, 2014, Little et al., 2008, Perrett et al., 1999, Rhodes, 2006, Thornhill and Gangestad, 1999) and may be an indicator of health (Thornhill and Gangestad, 1999). Findings are similar for sexually dimorphic faces (females with feminine features and males with masculine features) (Grammer and Thornhill, 1994, Griffey and Little, 2014, Little et al., 2008, Rhodes, 2006) and for symmetrical bodies (Concar, 1995, Ganstead and Simpson, 2000). Symmetry is positively correlated with number of lifetime sexual partners and a negative correlation has been found between asymmetry and age of first sexual intercourse in men only (Thornhill and Gangestad, 1994). This explanation does not however, explain why fewer women in my study had difficulties in this area, but were equally affected by asymmetry.
Gynaecological problems may have led some female participants to feel they fallen short of the feminine ideal, as well as contributing to psychological distress. Participants in my study were told about their gynaecological problems and fertility issues when they were teenagers. Infertility in women with Turner syndrome was found to have a greater impact than height in a qualitative study investigating challenges of living with Turner syndrome across the life span (Sutton et al., 2005). Sutton et al. described infertility as a continually sensitive issue throughout the life span, with participants being reminded every time a friend had a baby or grandchild or it was Mother’s Day. One participant said she felt “incomplete” (pg. 59) due to not being able to have children. Research into infertility and gynaecological problems report similar findings; women report feeling like girls and not grown women, that they did not feel ‘feminine’ or ‘normal’ and that they were inadequate and a failure (Kitzinger, 2002).

Challenges during adolescence became even more salient for participants as they were visibly different and were living with a health condition, as some of them viewed it. Models of social anxiety and stigma provide a framework with which to understand these challenges. Bullying and stigmatisation was viewed as confirmation participants were different. They had health professional’s regularly measure their height, parents worry about whether they will grow taller and what future they will have, and teachers were advising their parents they would perform better in a special school; all this must have influenced their views of themselves. They described a cycle of experiences that may have started with one incident or an accumulation of issues, which slowly eroded their confidence. Participants began avoiding interacting with others and their world started to shrink. They were anxious about how others perceived them and these anxieties were reflected back through others’ stigmatising or discriminating behaviour towards them.

Others may have believed that participants had the mental and physical capabilities equal to their size, and in turn did not possess the type of thoughts and feelings affected by negative treatment (Gollust et al., 2003). People perceiving the person they have stigmatised because of their stature, asymmetry or other physical characteristics, do so in a way as to dehumanise them (Goffman, 1963). This perception would have allowed them to bully, stare or make comments without feeling it was morally apprehensible. Participants felt they were being compared unfavourably to others and began to
withdraw from social interactions. This meant that they might have missed out on developing social skills, which make future interactions more awkward and less likely to develop into long-term connections. This re-confirms that fact that they have fallen short in some way. Participants with Achondroplasia also reported feeling stigmatised (Gollust et al., 2003). The pressure of trying to conform and the fear of bullying and stigmatisation may have made them reluctant to identify with their condition and others with RSS, therefore not feeling able to access support groups and other forms of support.

Contrary to previous literature, the majority of participants reported no issues with developing friendships. For example, the literature reports that appearance has an effect on friendships, as children and adolescents prefer attractive peers to play and work with in school (Boyatziz et al., 1998, Dion, 1973, Langlois and Downs, 1979). This preference extended to attributing more positive characteristics to attractive people and preferring them as friends in adulthood (Lemay et al., 2010). The participants in this sample reported being adept at making friends and described themselves as popular. One explanation for this incongruence with the previous literature may lie with the ‘what is beautiful is good’ relationship being reversible (Gross and Crofton, 1977). The more time we spend with someone and the more we grow to like and value them, the more attractive they become. This is significant when applying this to romantic relationships where positive characteristics are more likely to be attributed to appearance rather than personality initially. Another possible explanation could be appearance issues for people with RSS are more about how they feel about themselves or that for men with RSS, appearance is more pertinent to romantic relationships than friendships.

5.1.2 “You don’t have Russell-Silver anymore; it’s only a childhood condition”

Though many participants described their experience of the healthcare system as positive and their visits to the children’s hospital with fondness, they reported some negative events and experiences; for example, participants did not describe a smooth transition of care between childhood and adult services. Their experiences of healthcare in adulthood were those of neglect, health anxiety, and of not being taken seriously by non-specialist health professionals who often labelled medical complaints under the “umbrella” diagnosis of RSS. This gave them the impression that, within the healthcare system, RSS was not a recognised condition affecting adults, and one participant was told, by a health professional, that they no longer had RSS at it was a childhood condition. However,
participants clearly had health issues and challenges as adults. These experiences are similar to those of people that have conditions that are medically unexplained, such as Chronic Fatigue syndrome, where people often feel that health professionals are unsympathetic and that they are not believed or taken seriously (Anderson et al., 2012, Picariello et al., 2015). Worryingly for these participants, experiencing unexplained symptoms can often have a greater impact on physical and psychological functioning than the same symptoms in a diagnosed condition (Moss-Morris and Chalder, 2003). Comparisons can also be drawn between RSS and conditions like cleft lip/palate. Unlike RSS the cleft service has been redesigned in the last decade to encompass adulthood (Stock et al., 2015). Some people with cleft lip/palate had poorer psychological adjustment, which may have reflected the lack of psychological support once offered in the service. Also, research has found that new issues arise in adulthood for people with cleft lip/palate – similar to those experienced by people with RSS - such as starting work and forming long-term romantic relationships, not addressed in the under 18 service, which could direct further support.

Unfortunately, recommendations for the care of adults with RSS were not included in the recent expert consensus statement (Wakeling et al., 2016) as the experts concluded that of the few cases of people with RSS being followed up in adulthood, none had experienced health issues. Healthcare currently covers a period between birth and reaching final height (between 14 and 16 years of age). My data shows that adults with RSS experience significant issues in adulthood yet specialist care ceased when participants were experiencing some of their most challenging times, for example, failure to form romantic relationships, being informed of infertility, starting college and finding work.

I made the case in the introduction for why the lived experience of RSS might not be adequately explained through theoretical concepts such as biographical disruption and that the literature on visible difference may provide a better framework based on the literature describing the experience of people with Achondroplasia, Turner syndrome and Constitutional Growth Delay. However, upon analysing the interview data and identifying the key finding it became apparent that – as well as the experience of living with a visible difference - the Common Sense Model of self-regulation of health and illness, which from
this point forward I will refer to as the Common Sense Model for brevity, helps understand these findings. The Common Sense Model (Leventhal et al., 2003) may help interpret cognitive and behavioural factors and individual differences in relation to coping with a chronic health condition. However, one aspect of Bury’s theory corresponds with my findings: the search for the cause of an illness was an aspect of experience of onset and subsequent development of chronic illness (Bury, 1982).

Illness cognitions are defined as common sense beliefs about illness; these beliefs then shape how a person copes and understands their illness (Ogden, 2007). Knowing this information can inform targeted interventions to improve coping (Petrie et al., 1996). There are five domains to the Common Sense Model (Leventhal et al., 2003). Walter and Emery (2006) qualitative study, found that experience of cancer was defined by outside factors such as family and friends having cancer (identity); that they perceived cancer to be an “inevitable death sentence” (Walter and Emery, 2006, pp. 475) (timeline); it was a “threatening and serious disease” (pp. 475) (consequence); participants thought there was more than one cause i.e. environmental and genetic (cause); and they perceived themselves to have more control over environmental factors than genetic (control). Stimuli generated by an illness are converted into representations, which may be perceived as threatening, leading to action and appraisal. The Common Sense Model has validity across a wide range of illnesses (Hagger and Orbell, 2003) although there are some limitations. It does not acknowledge a person’s expectations of health services and how past experiences of health services contribute to current illness representations and also emotions are “inadequately defined” (Wyke et al., 2013, pp. 82).

Table 6 below details the five domains of lay illness representations illustrated using quotes from one participant, Bonnie. Bonnie appeared hyper-vigilant regarding any new health issues and coped with this through frequent visits to her GP. In Bonnie’s case her GP understood and was helpful though this was not the case for other participants. The absence of information regarding RSS in adulthood left Bonnie’s GP unable to confidently diagnose her health issues as related to RSS, which left Bonnie anxious that the cause may be more sinister. She started to make changes such as reducing her hours at work and spending more time resting due to fearing her health would deteriorate further. Having a label for her symptoms may have helped Bonnie to move from appraising the
situation as threatening (impotent to move forward) to appraising it as more of a challenge (taking steps to manage her health). The emotional cost of this health behaviour was that she began to feel less in control and became more reliant on her husband and children.

Table 7. The five domains of illness representations illustrated with participant (Bonnie) quotes (Leventhal et al., 2003).

<table>
<thead>
<tr>
<th>Domain</th>
<th>Description</th>
</tr>
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<tbody>
<tr>
<td>IDENTITY (label)</td>
<td>Participants reported various symptoms in adulthood, such as, fatigue, joint pain, weight gain, poor immune system, diabetes, and headaches: “Yeah, I get headaches, you know if you get that tired, you know when you get that tired you feel that worn out and then you get a headache and you feel sick, I’ve just got to go to bed.”</td>
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<tr>
<td>TIMELINE (duration)</td>
<td>Participants were often under the impression that they would no longer be affected by RSS in adulthood or that their symptoms would remain stable: “Because obviously child growth [Child Growth Foundation], like you think children, but then when I looked into it I found out there was adults, and I just rang up […] also as well to find out if other people were having these same problems.”</td>
</tr>
<tr>
<td>CONSEQUENCES (expected outcomes)</td>
<td>Participants described feeling uncomfortable with the unknown aspect of how RSS would affect them in adulthood. New symptoms - or the persistence of existing symptoms – could not be unequivocally attributed to RSS, which resulted in anxiety: “I think if somebody said to me having the low pressure and the chronic fatigue and things, that is typical of people with Russell Silver, then I can just think yeah, there’s no underlying cause […] because you start to think ‘Oh could it be something else?’”</td>
</tr>
<tr>
<td>CAUSE</td>
<td>Specialist and non-specialist health professionals could not attribute a cause to the various symptoms experienced in adulthood: “I don’t know, because the thing is when they look into things they’ll [GP] just turn around and say ‘We don’t know, so we’ll put it down to the Russell Silver because we don’t really know enough about that.’”</td>
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<tr>
<td>CONTROL (yes/no)</td>
<td>As treatment for RSS was isolated to childhood participants felt neglected by the health care system in adulthood. Control was lost and many questions remained about health outcomes and diagnosis and treatment for adult symptoms: “Has it been found out that no it’s not Russell Silver but it could be linked to that, and (pause) you know maybe if there’s alternative treatments.”</td>
</tr>
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Aspects of the Common Sense Model provide some understanding of why no care provision exists for adults with RSS. Lynch et al. (2015) used the Common Sense Model to
highlight a disparity between health professionals’ and patients’ beliefs about the length of a depressive episode. This disparity led to anxiety in patients when advised to stop treatment, as they believed it was too soon. Similarly, my findings show that participants had anxieties about not necessarily stopping growth hormone treatment, but certainly not having regular appointments with a specialist who also knew their medical history. If health professionals treat RSS as a childhood condition, this then dictates the timeline in which this condition will be managed, even if this is opposed to the patient’s timeline. Health professionals know this is a lifelong condition, but research does now exist – in the form of my study - to inform what the lived experience is in adulthood, but the healthcare system is not set up for adults with RSS. Aspects, such as timeline and consequences, were unknowns for participants and health professionals, which may have contributed to RSS being seen as a childhood condition. With participants, their families, and health professionals treating RSS in this way there was no need to acknowledge the possibility of issues in adulthood. This may have resulted in participants feeling confused about the aetiology of their adult issues. Believing that your condition will disappear in adulthood may have been a way of coping with having a health condition. Consequently, issues experienced in adulthood are harder to cope with as participants were unsure at times which symptoms were related to RSS and which were not. The Common Sense Model does not account for the influence or impact of friends, family or health professionals’ illness representations in regard to the person with RSS and the impact these have the on participant’s illness representations. It would be expected that family illness representations will influence and shape children’s illness representations in cases of congenital conditions like RSS.

5.1.3 A mismatch between healthcare provision and patient need
One characteristic that sets people with RSS apart from others is their short stature. However, the results from this study show that participants were psychosocially affected by appearance-related concerns above and beyond height. Participants gave many examples where their height was measured and plotted on a graph during hospital visits, they discussed having growth hormone injections every day and going to conventions where parents would discuss with other parents their hopes for their child to grow taller. However, height was not participants’ main concern. This is noteworthy as the main treatment for RSS is growth hormone therapy to gain height. This points towards a
mismatch between the desired outcome for health professionals and the needs of the patient. Through discussions with an endocrinologist, clinical geneticist and paediatrician, attention is concentrated on height in any contact with a health professional for patients with RSS. These results challenge previously held beliefs by health professionals working with patients with RSS by illustrating that this group of people potentially have many other concerns.

It is possible that the nature and focus of hospital visits may also affect participants adversely. Zlotkin and Varma (2006) found that children being treated for growth hormone deficiency (which meant a greater number of medical consultations) had greater psychosocial issues than those with idiopathic short stature. My findings resonate more closely to those of other short stature syndromes, such as Achondroplasia and Turner syndrome, compared with studies exploring the experience of people with idiopathic short stature or people with no medical cause for their short stature. It is possible that because participants perceived height to be health professionals’ main focus they internalised this and began to focus on their appearance, which may have increased psychological distress and affected self-esteem. This may have led participants to believe they could not discuss other issues with their health professional as their appointments were focused around growth, which in turn exacerbates feelings of being different. This would make it challenging for health professionals to identify psychological distress.

I have outlined the psychosocial issues participants experienced during adolescence. Those that talked about social avoidance were also the participants that talked about their appearance particularly negatively, had experienced negative reactions from others, and also self-harmed. For appearance-related concerns, social avoidance is more often associated with distress (Moss and Rosser, 2012). The psychological distress experienced by participants is an important finding, as not only did it appear to affect aspects of participant’s lives, but it is also set against the backdrop of healthcare provision for this patient group. Significantly, no routine psychological support is offered to patients with RSS and contact with specialist health professionals ceases when final height is reached.5

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5 With the exception of one participant who has a yearly appointment with an adult endocrinologist. The reasons for this were not clear from the interview with this participant.
There does not appear to be concordance between patients and health professional’s health and illness beliefs for RSS. If patient’s beliefs are not aligned to health professional’s beliefs this could result in lower patient satisfaction with care (Petrie et al., 1996). Participants may have felt they had some control over their condition when using growth hormone therapy, but this sense of control diminishes once they reach their final height.

Lack of healthcare provision in adulthood may have directed participants to seek support from face-to-face and online groups, which may not have been as helpful as support from a health professional. Though there have been many studies reporting on the benefits of accessing support groups: people gain social capital from the relationships they form with others in a similar situation (Clark et al., 2013), downward social comparison in a support group setting can enhance self-esteem (Finlay and Elander, 2016), upward social comparison can foster hope and inspire people (Carmack Taylor et al., 2007, Proudfoot et al., 2012, Vilhauer, 2011), and they can alleviate feelings of being different (Carmack Taylor et al., 2007, Finlay and Elander, 2016). Some participants did find support groups beneficial and for others the support they were hoping for was illusive. Due to the heterogeneous nature of RSS, participants found it hard to identify and empathise with others with the condition. Some felt like a fraud or an imposter within the group, which was also reported in a study exploring eating disorder support groups and led those participants to break contact with the group (Gale et al., 2016). Also RSS is not something you will recover from, participants would not have benefited from seeing others better off than them.

5.1.4 What patients, families and health professional need to know about living with RSS

I did not detect a discernible difference between those that had growth hormone treatment and those that did not. This finding should be reported with caution, as I am not stating here that families should not consider treatment with growth hormone therapy, rather the idea that better conversations can be had between health professionals and families about treatments and the management of expectations. The biggest difference found was between men and women and their experiences in forming romantic relationships. Men had greater appearance-related concerns, and some participants were notably affected by their asymmetry, which growth hormone does not treat.
My research shows the importance of people with RSS being encouraged to build strong friendships. Relationships with friends, family, and partners can have a positive effect on resilience through supportive mechanisms that enhance self-esteem, enable the development of social skills and provide a sense of belonging (Rumsey and Harcourt, 2012). Though most people I interviewed had strong friendships, there were a few that did not. Support to enable friendship building may have been beneficial for this minority. Being encouraged by family and efforts made by the individual to capitalise on social interaction and strengthening social relationships could increase resilience against adversity resulting from visible differences (Feragen et al., 2010). The downside is that these relationships can also threaten resilience through bullying and upward social comparison (Rumsey and Harcourt, 2012).

The present study, without this being the main focus of discussion with participants, identified adaptive ways of coping with adversity. This is noteworthy as it must be heartening for patients and families of those with RSS to know that people with RSS can be, and are, resilient. Resilience is the ability to recover from adverse events; adversity could be a single event, but for those with a congenital condition or visible difference this could affect the individual at multiple times across their life span. Participants also found positives of living with RSS, made downward comparisons, and valued other characteristics over appearance, which have been identified as an adaptive way of coping in other conditions (Baker et al., 2009, Egan et al., 2011).

Those with co-morbidities may have been more at risk of psychological distress. The participants that had the most significant issues were the ones with additional health conditions, echoing findings by Feragen and Stock (2014). They identified participants with cleft lip/palate and other co-morbid conditions may be more likely to have poorer psychological health and poorer satisfaction with relationships. Adolescence is a difficult time for any teenager; having appearance-related concerns and a congenital health condition may compound normal adolescent issues further. However, it is important to add that at the time point I interviewed this sample, despite past challenges, they were mostly at a positive point in their lives. They described successful and satisfying careers, happy relationships and some had started their own families.
5.2 Recommendations

In light of my findings, I have made several recommendations below. These recommendations cannot address all the issues reported in this study as similarly not every person with RSS would relate to all or any of the issues participants have experienced. However, bringing these issues to the attention of healthcare providers and commissioners will lay the foundations for making changes in attitude and practice for young people and adults with RSS.

5.2.1 Psychosocial issues and concerns other than height

Participants reported psychosocial issues in adolescence not addressed by health professionals. If these issues were recognised early enough and treated appropriately, it is possible the impact and duration could have been reduced. Health professionals working with children with RSS may understandably focus on physical characteristics where effective treatment exists. My research highlights a tension between focusing on height, as that is where a significant difference can be made, and the significance of whether reaching final height ameliorates psychosocial issues. This focus on final height means that consultations should not be structured in a way that inhibits the elucidation of psychosocial issues. It must be stated here that there are significant benefits for children with RSS to be treated with growth hormone therapy other than gaining height, for example, to increase muscle mass, muscle function and appetite (Schweizer et al., 2008, Willemsen et al., 2007), though evidence suggests growth hormone therapy does not improve psychosocial issues (Balen et al., 2006). Also a recent systematic review has concluded that studies claiming growth hormone ameliorates psychological distress are of poor quality and methodologically flawed (Gardner et al., 2016). Interestingly, although many participants overtly discussed psychological distress, some stated RSS or artifacts of having RSS, such as staring and comments, had no impact. However, further questioning provided evidence to the contrary, as they described issues with their appearance, anxiety and depression. It may be the case that they truly were not affected, but if there is a small chance their quality of life could be improved by a more person-centered approach, then psychosocial assessment and intervention is applicable to a larger number than my study indicates.
I would recommend, as an adjunct to treatment and clinical guidance, health professionals become more vigilant in identifying appearance-related concerns and psychosocial issues in people with RSS. This guidance could be extended to other conditions that affect growth as research has found similar challenges in Achondroplasia and Turner syndrome (Boman et al., 1998, Carel et al., 2006, Gollust et al., 2003, Kagan-Krieger, 1998, Sutton et al., 2005). Even though not all patients need extra support due to individual differences in coping and adjusting to short stature (Cohen et al., 2008, Noeker et al., 2012), and visible difference (Noeker et al., 2012), all patients should be screened for psychosocial issues. In the most recent recommendations for the management of RSS (Wakeling et al., 2016) the experts “recommend” psychosocial assessment at diagnosis. Psychosocial assessment at aged 2-10 and 10-18 is termed as “consider assessment, depending on the clinical features” (pp. 16). This is concerning based on the visible difference research that shows psychological distress does not related to objective severity (Rumsey and Harcourt, 2005b). I would argue, based on the findings from this study, that psychosocial assessment should be escalated to “recommended” and that the findings from my study are combined with the expert consensus report to inform a care pathway for young people and adults with RSS.

Those involved in the care of people with RSS could also look to incorporate patient reported outcome measures (PROMs). PROMs “seek to ascertain patients’ views of their symptoms, their functional status, and their health related quality of life” (Black, 2013, pp. 1) to improve care for individual patients. The measure can also be tailored to specific conditions and would need adapting for use in adolescents. Using PROMs may reduce the mismatch between health professional’s focus on height and patients concerns above and beyond height. These recommendations are similar to research by others, for example, Balen et al. (2006) who recommended health professionals focus on factors other than height to improve psychosocial adjustment in children and adolescence with idiopathic short stature.

5.2.2 Psychosocial interventions

Forming romantic relationships was a distinct challenge for male participants. This issue not only spanned adolescence and adulthood, but forming romantic relationships in adolescence is an important source of self-worth (Sanchez et al., 2008); missing out this stage may have important implications for social and psychological functioning in
adulthood. Also, the length of time participants had to live with this issue had implications. Recent research shows that single men feel less physically attractive compared with single women; those that have been single for over five years feel the least attractive, single women did not differ in feelings of attractiveness from women in general (Ochnik and Mandal, 2016). This indicates that the issue for men is exacerbated the longer they remain single. This challenge, along with its appearance-related antecedents, supports a strong argument for the provision of psychosocial support for people with RSS.

When reflecting on his experiences of the healthcare system, one participant thought health professionals (and families) focused too much on height when they should have focused on building self-esteem. He expressed the view that psychological interventions were just as important, if not more important, than medical interventions. RSS cannot be cured and there is a limit to the impact medical interventions, such as growth hormone therapy, can have. Well-timed evidence-based interventions could enhance resilience, confidence and self-esteem. Figure 5 shows how the experience of having RSS changes from early childhood to early adulthood and where interventions could be targeted.

In a systematic review of psychosocial interventions for young people with visible differences, Jenkinson et al. (2015) found limited evidence of effectiveness for social interactive skills training, cognitive behavioural therapy and education/behavioral therapy. These interventions generally aimed to improve self-esteem, self-confidence, difficulties with social interactions, poor body image, and behavioural issues (ibid). The current guidelines for the management of RSS recommend providing information about support groups at diagnosis and not at any other stage (Wakeling et al., 2016). However, considering the findings of this study, and evidence from appearance psychology, health professionals could initially consider providing low-level interventions as suggested by the Centre for Appearance (CAR) framework of interventions for people with visible differences (Jenkinson, 2012), which include information giving and directing patients towards support groups (face-to-face and online).

However, the heterogeneous nature of RSS made it difficult for some to find commonalities with others with RSS, whilst also feeling different to everyone else. This
resulted in support becoming illusive for participants. Though it could be argued that participants may benefit from downward social comparisons through accessing support groups, what if you are the worst affected person?\(^6\) This leads me to conclude that support groups may be an ‘easy option’ for specialist and non-specialist health professionals to advise people with RSS to attend, and my findings show that this must not be the only support offered. Case studies of others with RSS could be used to manage expectations in adolescence and adulthood. Having vignettes, assembled from the participant data I have collected, may ‘bring to life’ what living with RSS is really like, enabling families and people with RSS to plan for any challenges and have more informed discussions with health professional about their care.

Some participants capitalised on characteristics other than appearance as a way of coping or gaining social capital. Moss and Carr (2004) suggested intervention techniques that target integrating non-appearance related information into the self-concept as a worthwhile strategy. Whilst exploring self-concept organisation and wellbeing to understand individual variation in adjusting to visible difference, Moss and Carr (2004) found that poorer adjusters organise appearance-related information in a different way. Those who valued appearance over other characteristics were more likely to rate their appearance negatively and perceived others would also. Leading on from this work Griffiths et al. (2012) showed that this process leads to lower self-esteem and confidence when regarding romantic relationships.

One difficulty in recommending psychosocial interventions for people with RSS is teasing apart the health condition from the visible difference issues. Are they distinct models that led to distinct outcomes, both needing interventions tailored to their particular needs? I suspect it is more complicated than that, that they are inextricably linked. However, my results and participant experiences strongly resonate with those from other visible difference studies. Luke described the need for psychological interventions for people with RSS:

\(^6\) At the Child Growth Foundation convention I met a family attending for the first time. Amongst other reasons, one aim of attending was to feel as though their daughters’ condition was not as severe as it could have been. However, they found that she was much more affected than any other child they saw that day.
Luke

I think sometimes it highlights issues that aren’t really there [...] when they talk about growth hormone it puts pressure on ‘Look how different you are’ rather than building up on the inner confidence [...] it should be more about being a big person in a little body and then you know who you are, what you stand for and how to cope with things really. What’s important is your own self-esteem [...] I think more emphasis should be put on that.
Figure 5. Challenges faced by people with RSS in childhood, adolescence and early adulthood with recommendations for when psychological intervention could be most useful.

- **Aged 0-10**
  - Feeling different, but that is neither good or bad
  - Start growth hormone therapy
  - Psychological intervention with young person and family

- **10-14**
  - Feeling different takes on a more negative aspect
  - Early puberty
  - Appearance related concerns
  - Bullying and/or comments from others
  - Psychological distress
  - Psychological intervention with young person

- **15-18**
  - Romantic relationships
  - Infertility
  - Growth stops
  - Starting college/work
  - Visits to specialist healthcare stop
  - Psychological distress

- **18+**
  - Striving to live independently
  - Difficulty forming romantic relationships
  - Starting university/work
  - Psychological distress
5.2.3 Transition between child and adult healthcare

Health professionals, especially paediatric endocrinologists, know RSS is a lifelong condition; however, service provision does not reflect this. Health care is compartmentalised into specialties and is dependent on the age range of patients. However, the person does not feel that way. To them their health is a continuum, which often makes it difficult to understand how healthcare is organised. Healthcare providers and commissioners may need to re-think how patients transition successfully from child to adult services. In a systematic review of studies exploring the impact of childhood chronic health conditions and the impact on family members, Waldboth et al. (2016) concluded that transitioning into adulthood was a significant challenge for the young person. The authors recommended greater support for the transition into adulthood to reduce the psychosocial impact.

It is clear that current care for adults with RSS is severely lacking and participants are not satisfied with the support they receive from non-specialist health professionals. It is beyond my knowledge of the healthcare system to make recommendations for an adult RSS service, but recognition of the psychosocial challenges people with RSS potentially face as young people and as adults would be a positive move forwards.

5.2.4 What can non-specialist health professionals offer?

The focus does not have to be just on specialist health professionals. Non-specialist health professionals are ideally placed to identify difficulties faced by young people and adults with RSS. Waldboth et al. (2016) suggested that practice nurses and GPs are well placed to monitor psychological wellbeing and co-ordinate inter-disciplinary support if needed. Participants were critical, but understanding, of general practitioners lack of knowledge about RSS. A simple intervention, such as the production of literature for participants to give to any professional, explaining the possible challenges an adult with RSS could face could fill the knowledge gap. Although, it is difficult to predict what impact having more knowledge about RSS will have for GPs, as there are currently no specialist services to refer patients to, GPs have the advantage of cutting across all the medical disciplines. This means that they can refer patients to specialists such as orthotics, dieticians, and for psychological support.
5.3 Strengths and limitations

The findings from this research are novel and make a contribution to the field of health psychology in 6 ways:

1. The literature on RSS mainly describes clinical features and is focused on children (Ballard et al., 2016, Wakeling et al., 2016); my research adds depth and insight into the lived experience of adults with RSS.

2. Research on body image and appearance-related concerns in men is still relatively rare (Parent et al., 2016, Thompson, 2012) even though research has found that men have more concerns over body image than has previously been assumed (Grogan and Richards, 2002). Men in this study were more affected by appearance-related concerns than women and reasons for this were explored, which adds to the rare but growing literature base.

3. Research exploring how appearance-related concerns impact people’s lives has neglected to thoroughly explore how it affects romantic relationships (Griffiths et al., 2012) significant to men in this sample.

4. Research on appearance-related concerns and visible differences has focused heavily on the negative consequences for participants. There have been few studies exploring how individuals have remained resilient despite these differences and have flourished socially (Feragen et al., 2010, Moss and Rosser, 2012, Thompson, 2012). The participants in this particular group did describe positive experiences and remaining resilient despite challenges they may have faced or still are facing.

5. The sample of participants included a range of ages, participants that have undergone a range of treatment options and a mix of female and male participants. This allowed me to capture a variety of experiences to answer the research question.
6. This study was underpinned by patient and public involvement throughout by working with a Child Growth Foundation representative, who has a child with RSS.

This study adds a fuller understanding to explain why romantic relationships are difficult, which will better inform how people with RSS can be supported. Studies looking at similar conditions, such as Achondroplasia (Gollust et al., 2003), have used survey data to explore how participants perceive their condition and their quality of life. Though this study found issues similar to those found in my study, for example, one of the disadvantages of having Achondroplasia reported was “Difficulties in personal relationships, dating and socializing” (pp. 454), there provide no detail beyond this statement.

Validity in this study was enhanced firstly by the evolution of the interview schedule. Initially I adapted the interview schedule based on a pilot interview and initial participant interviews to reflect relevant topics discussed. I identified noteworthy themes in the data after starting the analysis and added extra questions to the schedule, for example, “If you could change one aspect of your RSS what would that be?” Secondly, participants talked candidly about their experiences and without prompting discussed personal issues, for example, Warren confided “it’s a bit awkward to say it, but it probably has affected me a little bit sexually as well. Erm in that I obviously am smaller than most guys down below”. And thirdly, I presented the key findings to adults with RSS and families of people with RSS as a way of triangulating my results. This gave me some reassurance that the conceptual work I had carried out was still rooted in participants’ experiences.

There were three main limitations to this study:

1. This research relied heavily on participants recalling past experiences, which could be influenced towards a higher reporting of memories from adolescence. This is called the reminiscence bump (Rubin and Schulkind, 1997), which describes adults recalling more autobiographical memories from the ages of between ten and thirty. This was however a pertinent time for people with RSS and is when treatment and regular hospital appointments ceased; it would be expected for adults to recall more experiences from this time. It would be prudent then, in a
future study, to interview adolescents in order to gain a fuller picture of the needs of young people with RSS.

2. The sample consisted of participants with two specific genetic types of RSS in an effort to gain homogeneity so may not be representative of, or applicable to, everyone with RSS. I have no reason to believe this sample is different to others with a clinical diagnosis, however exploring the lived experience of other types of RSS would add to the understanding gained from this study. Participants are those that came forward for the medical arm of the study. They may represent people with more complex issues who were keen to have a thorough medical examination or people who were able to make the trip to Southampton; others with more disabilities may have been unable to take part. Reflecting on the sample of participants I interviewed I would say this was not the case. These participants had a broad range of issues, some appeared to be almost unaffected, some had complex health issues and some classed themselves as disabled.

3. Teasing out which experiences were related to RSS and which were related to other health conditions or to personality factors was impossible. Other health conditions (psychological and physical) in addition to RSS, such as dyslexia, cleft lip/palate, and Asperger’s syndrome which could lead them to be more vulnerable to poor psychological adjustment (Feragen and Stock, 2014). What I have presented here is what participants believed to be related to their condition, interpretations I have made from the data and I have identified where these findings are similar or different to research on other conditions; these are the multiple truths of what it is like to live with RSS.

5.4 Future directions
These key findings need to be disseminated to people with RSS, families, healthcare commissioners, and health professionals involved in their care. Adults and families asked for child friendly resources, online and hard copies of information about the lived experience of RSS for other professionals such as teachers and mental health specialists. It may be possible to capitalise on a resource that has been developed called ‘My Medical Record’, an online health record that patients have access to and a way of sharing
information with other health professionals. It would be possible for a person with RSS to have information about what it is like to live with RSS on their record and make that available to people involved in their care. There is also a need for better quality studies investigating interventions to improve psychosocial adjustment in young people with visible differences.

There are other conditions that effect growth, such as Beckwith-Wiedemann Syndrome, which lack research exploring the lived experience. There is every chance similar issues will be found with other under researched conditions and the findings and recommendations from my study may be applicable to a wider group of patients in paediatric endocrinology clinics.

5.5 Conclusion
This study has highlighted that people with RSS may experience psychosocial issues in adolescence and adulthood resulting from appearance-related concerns, which overshadow a concern about height. These findings are noteworthy as specialist healthcare for people with RSS is often only provided until late adolescence and is focused on gaining height. Currently no psychological support is routinely offered to this patient group and no specialist service exists for adults to access. The findings from this study need to be considered alongside the most recent expert consensus statement for the treatment of RSS in order to inform a care pathway that extends into adulthood and incorporates recommendations for psychosocial assessment, intervention, and patient reported outcome measures. Health psychologists are therefore well placed to collaborate with clinicians to raise awareness of appearance-related concerns and ameliorate psychological distress in this overlooked patient group.
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Appendix 1 – Research diary extract

16/10/2016
I am just finishing off verifying and coding the adolescent transcripts. I have noticed another commonality in among women. They talk about falling over and loosing their balance. Corine, Temple syndrome, one that had leg surgery, any others? Three at least have mentioned headaches or migraines.

02/10/2016
I have three weeks until my final draft has to be with my supervisors so I have worked out a plan to get it written. Week one is concentrated on results.

[Sections of this research diary have been removed for brevity]

11/05/2016
Coding the rest of the transcripts
Mina and Oli both mention self-harming and talk very negatively about their appearance
Added a code for bulling, psychological distress - looked through other transcripts and searched for anxiety, anxious, sad, depression, upset, fear, worry, social, friend.
Appearance and body image
- Acceptance of how you look comes with age
- Being with peers makes you realise how different you are
- Bise I don’t like
- How I see myself isn’t how others see me - skewed perception
- I’d have rejected me if I was myself

1. Sexual Relationships
- Appearance and body image
- Compensating characteristics or being small helps
- Confidence or self esteem
- Feminine Ideal
- Masculine Ideal
- ‘Mayor of the friend zone’
- Reaction or impact of others (real or perceived)
- Accepting the reality (or not)
- Activities that overcome
- Being held back
- Being or feeling different
- ‘Being short is not the worlds worst thing’
- Chronic illness or pain
- Crossing the Rubicon (the point of no return)
- Disability
- Fitting in (with society or with rss)
- GH
- ‘Having the knowledge doesn’t make it better or worse for me’
- How I feel about having RSS
- How I talk to others about having RSS
- Keeping it to yourself
- Leg lengthening surgery
- Life span
- Over estimation of height
- Passing
- RSS or not RSS
- ‘RSS was slowing me down from growing up’
- Side Show
- Smoke and mirrors
- Social Comparison
- Standing on the sidelines
- Stigma (felt and enacted)
- Support (family, friends groups) (chimera - illusive)
- ‘Symmetry’s a good thing’
- Things health professionals say or do
- Transition
- Transition moments
- Uncertain future
- What group do I belong to
- XXXIdentity or labelling (diagnosis, passing, being something you’re

[Sections of this research diary have been removed for brevity]
17/03/2016
Reflection on interview number 17:

This was a tough interview to manage as the participant would really go off track and discuss things in great detail, I was mindful that there was a lot of description, but not much emotion or feeling. His wife was there and that was a blessing, as she seemed to really get what I wanted to know and would speed him up or quickly finish what he was saying to get him to move on. It was a slightly disturbing interview also as the participant seemed to have undergone a certain amount of abuse as an inpatient when he was a child. This was clearly something the participant wanted to convey and I did not hurry it along when he was relaying this part of his story. This participant was the oldest I have interviewed at 69, but he could still recall having trouble with getting a girl friend when he was young and relying on his sense of humour to win people round.

16/03/2016
Should I specify H19 and MatUPD7?

[Sections of this research diary have been removed for brevity]

04/03/2016
Going to go through the bigger codes to see if they themes themselves or need to be broken down into smaller codes.

Do I have some overarching themes? General (+ lifespan), male and female?

[Sections of this research diary have been removed for brevity]

04/02/2016
Luke 'it should be more about being a big person in a little body' - describes confidence and self-esteem that isn't derived from height or body image?
Should I be noting what participants say about their parent's experiences?
Luke 'brain tumour' health anxiety - what code does this go under?

02/02/2016
It has surprised me how quickly I can code the transcripts. It takes maybe 2-3 hours per transcript.

Where does bullying fit in? 'Reaction or impact of others (real or perceived)'

Being small and skinny is not as bad as being fat as far as bullying goes.

01/02/2016
Below is where my codes are now. I have been coding some more today and am going to add stigma (perceived) as my third transcript has just mentioned this, the first two didn't really and it's not something common that has come out of the interviews.
05/01/2016
Interview 15

The interview went well, the participant seemed at ease and discussed issues relating to RSS in depth without much prompting from me. He was relatively unaffected by RSS, but still brought up some interesting points. He kept his diagnosis quite private and mentioned why being smaller may have advantages for the kind of job he does, which got me thinking about other participants who had similar things. He did get a bit upset at the end when I asked him what motivated him to take part in this research, but it was more that he was grateful for the support he had received from HCPs rather than being disturbed by anything we had discussed. I made him aware that I was a bit concerned and that I would like to leave him as I found him and he reassured me that he was fine.

He had a question about the last item on the consent form, which I dealt with once I got back to the office and emailed him with the clarification.

[Sections of this research diary have been removed for brevity]

Interview 12

This was the second participant I have interviewed with matUPD7. It was a much easier interview, as the participant was really happy to share her story; earlier in the day when we arranged a time she said 'I've got so much to tell you'. This participant has learning difficulties, dyspraxia and traits of autism, which are common with matUPD7. It felt like a free flowing story and the participant got to say what she thought was relevant, I asked very few questions. This balances my male and female numbers and I had another person from the conference really interested, she says she will email me next week.

[Sections of this research diary have been removed for brevity]
Similarities - another self harming, also spoke about being embarrassed of RSS. Differences - was told she cannot have children

[Sections of this research diary have been removed for brevity]

19/10/2015
Changes to Interview Schedule
Made some changes to the interview schedule now that I have started to analyses the data I have so far and have some developing themes.
Removed
How did you find out - mostly they are too young
What has been the effect on your live - asked else where
Feeding problems and relationship with food now - not getting data that is different to what Kemi is already asking
Added
Has RSS affected how you meet potential romantic partners or affected your relationships in any way?
What aspect of your RSS has or had the biggest impact on your life?

[Sections of this research diary have been removed for brevity]

07/10/2015
Presented teaching session to clinical genetics team. It went well, I managed to make everyone laugh a bit as well, which I think helped (going back to my observed teaching by Charles Abraham). There weren't many questions after, but several members of the team did come up to me once I was back at my desk to compliment me on the talk.

[Sections of this research diary have been removed for brevity]

29/09/2015
More basic analysis. Took all the developing themes and set them out on paper. Printed out two transcripts on different coloured paper and cut out quotes that illustrate the themes. I will then start to explore each theme in more detail. Took a whole day!

[Sections of this research diary have been removed for brevity]

17/08/2015
Reflection on Interview 10
This interview was challenging as I had met the respondent before with Kemi and knew that his speech was quiet affected by his cleft palate. My main concern was that the audio recording wouldn't pick up what he was saying as in the previous medical appointment it was hard to hear what he was saying when you weren't looking at his mouth as well. I emailed a UWE researcher who does research on cleft palate and she has not had this issue before, but gave me some advise. One idea was to video record the interview to aid with transcription, but we couldn't source one in time and it would have meant an ethics amendment, which no one was keen on doing. I decided to audio record whilst also taking notes. I advised the respondent of this before we started the interview and talked about the fact that I would be taking notes and to carry on chatting and try not to feel that I wasn't listening. In reality this worked really well and I found that once we had been talking a while my ear 'tuned in' to his speech and I could hear him as I was writing. I haven't started transcribing yet though, so I still need to find out what the end result would be.

I really felt that this respondent challenged some pre-conceived ideas I had about him and what kind of life he would have. By the end of the interview I found his story inspiring. He has overcome severe confidence issues and has worked hard to get to where he is now and seems to be living a happy and fulfilling life.

[Sections of this research diary have been removed for brevity]

13/07/2015
This interview was quite different from the others as it was via Skype. As the connection was quite bad we turned off the camera and conducted it like a telephone call. The main difficulty with this was that I found it hard to know if the respondent had finished speaking or if they were leaving a pause, without non-verbal queues this is not easy to determine. The connection also broke completely three times during the interview. This didn't seem to bother the respondent, he was happy to pick up from where I could last hear him and chatted away confidently. I found it more of a challenge and brought the interview to a close quicker than I think I would have done if it were face to face. If I do another Skype interview I would consider doing it from home, where I know the Internet connection is reliable. I would like to do more interviews via Skype, especially as tomorrow I have another interview to conduct which will involve a 7 hour round trip in the car.

This respondent did not seem affected by RSS in adulthood and also had a different experience of intimate relationships in his teenage years compared with other male respondents.

13/7/2015
My supervisor doesn't like the term life span 'how does the lived experience of having RSS change across the life span' I have spent some time thinking of alternatives and I think it just comes down to sociologists who use life course and psychology using lifespan. I am going to keep lifespan, but what does it mean? Does it need defining?


17/06/2015

107
Reflection on interview six

I feel like it went well, the respondent was easy to talk to. It was clear from quite early on that he is pretty much unaffected by RSS. We mostly focused on height, being slight in build and dyslexia, which he wasn't sure was related to RSS. Another theme that has been repeated in other respondents came up again, which was lack of confidence socially. I asked him to give me an example a few times but he struggled to do this.
Appendix 2 – Cover letter

[Date]

[Patient’s name and address]

Dear [Patient’s name],

Re: Study of Adults and Adolescents with Russell-Silver Syndrome in the UK (STAARS UK)

You very kindly took part in the STAARS study and gave your consent to be contacted about future research.

I would now like to invite you to participate in the interview part of the study to find out more about what it means to have Russell Silver syndrome. This will take around one to two hours and I would hold the interview at a venue of your choice, like your home or local hospital.

I have included a copy of the information sheet. If you are interested in taking part, or have any questions, please contact me or the study team using the details on the sheet or return the response form.

Yours sincerely,

Lisa Ballard
Research Assistant
Appendix 3 – Participant information sheet

University of the West of England

Study of Adults and Adolescents with Russell Silver syndrome in the UK (STAARS UK)

INFORMATION BOOKLET FOR IN-DEPTH INTERVIEWS

For participants and parents/guardians

Contact the research team to take part in this study

REC reference number: 13/SC/0630
Version 1.2
Date: 11/03/15
You or your child have already taken part in research into Russell Silver syndrome. Thank you for letting us know that you would be happy to be involved further. We would now like to invite you to participate in the interview part of the study. This leaflet explains why we are doing this and what it would involve. If you have any questions, please contact Lisa Ballard, whose details can be found at the end of the sheet.

Purpose of the study
This is a study to find out more about what it means to have Russell Silver syndrome (RSS), a genetic condition affecting growth. We want to understand why it happens and whether there are long term health issues. In this part of our research, we want to understand what it is like to live with RSS and how the condition affects people’s lives.

Why have I been invited to take part?
You have kindly participated in the study already and indicated that you would be willing to be contacted again.

Do I have to take part?
No. It is up to you whether you take part. If you agree to take part and later change your mind, you can withdraw from the study. You can do this at any time. We will destroy your data if you want us to.

What will happen if I take part?
A member of our research team will interview you. This will take around 2 hours. We would hold the interview at a venue of your choice, like your home or local hospital. We will ask you about your life and experiences for example when you were growing up, your social life, school, work and how you think RSS has affected aspects of your life. With your permission, we will audio-record the interview, and later write up the recording. Your interview will help us to identify important issues about what it is like for people who have RSS.

What are the possible disadvantages and risks of taking part?
Talking about life experiences can sometimes be difficult or upsetting. If you find the interview difficult, you can stop and carry on at another time. Alternatively, you can withdraw from the study completely.
If you feel that you need further support with regard to any issues - either during or after the interview - please let your interviewer know and they will provide you with contact details for relevant support groups. You can also contact your GP or the clinical genetics team if you would like to talk to a healthcare professional about anything that comes up in the interview.
What are the possible benefits of taking part?
You may find it helpful to discuss your experiences. We will use the research findings to help healthcare professionals and families understand what it is like to live with RSS.

Will my taking part in the study be kept private and confidential?
Your data will be kept confidential—only the research team will see it.

We will store all your data securely. We will keep your name and contact details separate to your interview transcript (the written copy of your interview). We will remove identifying names or places from the transcript so that your identity is not known to readers of it.

In our reports or publications, we will include things that people say in their interviews. We will always make this information anonymous—we will never use a person’s own name. If there is any data that will clearly identify an individual, we will not use it in reports or publications.

We may use your interview transcript for future research. If you do not want us to, you can just let us know.

What if there is a problem?
If you have any other concerns or complaints about the study, you can contact the Patient Advice Liaison Service (PALS) for advice on 023 8120 8498.

What will happen to the results of the research study?
We will publish the findings of the research in journals for people working in universities and in healthcare. We have also set up a research project website (http://www.southampton.ac.uk/geneticimprinting/staars.page) where you can read about the research. You can contact us or refer to the website if you would like a summary of the main findings. If you would like the link to this website to be emailed to you, provide your email address to the researcher after your interview.

Who is organising and funding the research?
The research team organising the study is Professor Karen Temple, Dr Kemi Sodipe, Dr Angela Fenwick, and Lisa Ballard. This research is funded by the National Institute for Health Research (NIHR). The study is organised by the University Hospital Southampton and the University of Southampton.

Who has reviewed the study?
An independent group of people, called a Research Ethics Committee, review all research in the NHS. A committee has reviewed and approved this study (ethics number 13/SC/0630).

What do I do now?
If you would like to take part in this interview stage of the study, please let us know using one of the methods below. One of our team will then contact you to discuss
this in more detail, and give you chance to ask questions. If you still want to take part, they will arrange a suitable time and venue for the interview. You will complete a different consent form before the interview and have another opportunity to ask any questions and can still withdraw from the study.

Further information
If you have any queries, please contact:
Lisa Ballard
Tel: 023 8120 4462 (office)
Tel: 023 8120 6551 (research office)
E-mail: lisa.ballard@uhs.nhs.uk
Thank you for taking the time to read this information.

Contact the STAARS team
1. Phone 02381 206551

2. E-mail: staars@uhs.nhs.uk

3. Return this section and send to:

Study of Adults and Adolescents with Russell Silver syndrome in the UK (STAARS UK):
Wessex Clinical Genetics Service
Princess Anne Hospital
Coxford Road
Southampton
SO16 5YA

Provide us with:
NAME
ADDRESS
PREFERRED CONTACT NUMBER
Appendix 4 – Interview schedule

Study of Adults and Adolescents with Russell Silver Syndrome in the UK (STAARS UK)
In-depth interview framework (V1.3)

Starting questions and prompts
Tell me about when were you first aware of having your condition?
What was that like?
What did you think it meant?
What did you notice?
How has this changed over time?
What did/does having a genetic condition mean to you?
What is it like now?
What do you know about RSS?

School
Where did you go to school/college/university?
What was school like?
Can you describe any friendships at school/outside of school?
What sort of activities were you interested in
Has RSS restricted your life in anyway (then/now) or what did/do you want to do but haven’t/can’t?
Similar exploration of college/university

Intimate relationships
Life/ friendships and intimate relationships after school and now.
Has RSS affected how you meet potential romantic partners or affected your relationships in any way?
Planning and having children?

Work
Tell us about your work now and from when you left school.

Discrimination
Can you think of a time when you experienced any discrimination or people were unkind about your height or any other aspect of RSS?
Can you describe that?
What was it like for you?
Has it had any longer term impact?

Adjustments and general life
Do you think you have had to make any adjustments in your life because of RSS? Explore these.
Today, how do you think RSS impacts on your life, health and illness?
How does RSS affect everyday life?
How do you tell the difference between your RSS symptoms and other health symptoms?
Do you feel you have to explain RSS when you meet new people or enter new environments? To what extent do you talk about the condition and to whom?

Healthcare
Has the healthcare system been a bigger part of your life compared with others around you?
What has your experience been regarding health care professionals (your doctor’s surgery, at the hospital etc) and your condition?

Growth Hormone related questions
Did you/do you take GH? Explore when started and for how long as appropriate.
Explore reasons if not taking.
Side effects?
Impact on life, health and illness?

Closing questions
What aspect of your RSS has or had the biggest impact on your life?
What are your worries or hopes for the future?
You have given us a considerable amount of your time between this session and the first.
What does it mean to you to participate in this research?
Appendix 5 – Interview schedule tracked changes

V 1.1

Study of Adults and Adolescents with Russell Silver Syndrome in the UK (STAARS UK)

In-depth interview framework

Starting questions and prompts

When were you first aware of having your condition? How did you find out? What was that like?

What did you think it meant? What did you notice? What has been the effect on your life? How has this changed over time? What did/does having a genetic condition mean to you? What is it like now?

Where did you go to school? What was school like? Can you describe any friendships at school/ outside of school? What sort of activities were you interested in and has RSS restricted your life in any way (then/ now) or what did/ do you want to do but haven’t/ can’t?

Similar exploration of college/university

Life/ friendships and intimate relationships after school and now. Planning and having children?

Do you work and if so what do you do? Tell us about your work now and from when you left school.

Can you think of a time when you experienced any discrimination or people were unkind about your height? Can you describe that? What was it like for you? Has it had any longer term impact?

Do you think you have had to make any adjustments in your life because of RSS? Explore these. Today, how do you think RSS impacts on your life, health and illness? How does RSS affect everyday life? How do you tell the difference between your RSS symptoms and other health symptoms?

In my experience, people with RSS often have a problem feeding as a child, how has this affected your relationship with food as an adult? Do you ‘enjoy’ food?

Do you feel you have to explain RSS when you meet new people or enter new environments? To what extent do you talk about the condition and to whom?

What has your experience been regarding healthcare professionals (your doctor’s surgery, at the hospital etc) and your condition?

 Growth Hormone related questions

Did you/ do you take GH? Explore when started and for how long as appropriate. Explore reasons if not taking. Side effects? Impact on life, health and illness?

Closing questions

What are your worries or hopes for the future?

You have given us a considerable amount of your time between this session and the first. What does it mean to you to participate in this research?

V 1.2
Study of Adults and Adolescents with Russell Silver Syndrome in the UK (STAARS UK)

In-depth interview framework

Starting questions and prompts

Tell me about when were you first aware of having your condition?
How did you find out?
Was that like?
What did you think it meant?
What did you notice?
What has been the effect on your life?
How has this changed over time?
What did/do you mean by an acute condition mean to you?
What is it like now?
What do you know about RSS?

School
Where did you go to school/college/university?
What was school like?
Can you describe any friendships at school/college/university?
What sort of activities were you interested in?
Was RSS restricted your life in anyway (then/now) or what did/do you want to do but haven’t/can’t?
Similar exploration of college/university

Intimate relationships
Life, friendships and intimate relationships after school and now.
Planning and having children?

Work
Do you work and if so what do you do?
Tell us about your work now and from when you left school?

Discrimination
Can you think of a time when you experienced any discrimination or were unkind about your height or any aspect of RSS?
Can you describe that?
What was it like for you?
Has it had any longer term impact?

Adjustments and general life
Do you think you have had to make any adjustments in your life because of RSS?
Explore these.
Today, how do you think RSS impacts on your life, health and illness?
How does RSS affect everyday life?
How do you tell the difference between your RSS symptoms and other health symptoms?

In my experience, people with RSS often have a problem feeding as a child, how has this affected your relationship with food as an adult?
Do you feel you have to explain RSS when you meet new people or enter new environments?
To what extent do you talk about the condition and to whom?

Healthcare
Has the healthcare system been bigger part of your life compared to others around you?
What has your experience been regarding healthcare professionals (your doctor’s surgery, at the hospital etc) and your condition?

Growth Hormone related questions
Did you/do you take GH?
Explore when started and for how long as appropriate.
Explore reasons if not taking.
Side effects?
Impact on life, health and illness?

Closing questions
What are your worries or hopes for the future?
You have given us a considerable amount of your time between this session and the first. What does it mean to you to participate in this research?
Study of Adults and Adolescents with Russell Silver Syndrome in the UK (STAARS UK)

In-depth interview framework

Starting questions and prompts
Tell me about when were you first aware of having your condition?
- How did you find out?
- What was that like?
- What did you think it meant?
- What did you notice?
- What has the condition affected on your life?
- How has this changed over time?
- What did does a genetic condition mean to you?
- What is it like now?
- What do you know about RSS?

School
Where did you go to school/college/university?
- What was school like?
- Can you describe any friendships at school/otherwise of school?
- What sort of activities were you interested in?
- Has RSS affected your life in anyway (then/now) or what did/don’t you want to do but haven’t/can’t?
- Similar exploration of college/university

Intimate relationships
Life/friendships and intimate relationships after school and now.
- Has RSS affected how you meet potential romantic partner or affected your relationships in any way?
- Planning and having children?

Work
Tell us about your work now and from when you left school.

Discrimination
Can you think of a time when you experienced any discrimination or people were unkind about your height or any other aspect of RSS?
- Can you describe that?
- What was it like for you?
- Has it had any longer term impact?

Adjustments and general life
Do you think you have had to make any adjustments in your life because of RSS? Explore these.
- Today, how do you think RSS impacts on your life, health and illness?
- How does RSS affect everyday life?

How do you tell the difference between your RSS symptoms and other health symptoms?
Appendix 6 – Development of the thematic map
Appendix 7 – Consent form

IN-DEPTH INTERVIEW CONSENT FORM

Participating adult

1. I confirm that I have read and understand the information sheet dated ______ about the above study. I have been given the opportunity to consider the information, ask questions and have had these answered fully.

2. I understand that my participation is voluntary and that I can withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

3. I understand that sound recording will be used to help with the accurate recall of my responses and that I can request that the equipment be switched off at any time during the interview.

4. I understand that my data collected during the study, may be looked at by the research team, by individuals from regulatory authorities, or from the NHS Trust, where it is relevant to my taking part in this research. I give permission for these individuals to have access to my records.

5. I give permission for some of my responses to be used in reports from the study provided there are no links to my name.

6. I agree to take part in the interview study

OPTIONAL

7. I give permission for my interview transcript to be used in future ethically approved studies related to growth or RSS provided that this is not linked to my name.

Name of participant ___________________ Signature ___________________ Date ___________________

Name of person taking consent ___________________ Signature ___________________ Date ___________________
Appendix 8 – Example transcript

INTERVIEWER
Okay. So kind of the way I’ve started off the conversation is just getting people to tell me a bit about when they first remember having Russell Silver and kind of just talk a little bit about that. How was that for you?

MINA_F_25_GH_4’8
I was really little. I don’t remember how little I was, but my earliest memory is my mum used to come in when I was asleep, to do my injections, that was with the old pen, and then I remember the lady coming round to do like my once a month injections. And then I remember we got a new pen, and I remember like practicing how to do it on oranges with my mum, and then she taught me like how to do it myself and everything like that.

INTERVIEWER
Okay. So these injections, were they your growth hormone injections?

MINA_F_25_GH_4’8
Yeah, yeah, which I found quite easy. I quite liked doing it, because it’s something that I was responsible for doing it, and it was quite like an important thing and I had to remember it and I knew how to put the needle on, take it off, change it and everything like that, so that was good.

INTERVIEWER
How old were you when you first started doing them yourself?

MINA_F_25_GH_4’8
I’m not too sure, I’d say probably about six or seven, at a guess.

INTERVIEWER
So quite young.

MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
Yeah. So your earliest memories are having your injections. So do you remember what, whether you knew why you were having those injections?

MINA_F_25_GH_4’8
I think I did on so you know because my mum was like, well because my mum and dad had separated, and they both took a different approach to it, which was a bit strange, but because I always lived my mum, so I didn’t mind injections and whatnot. My mum was like they’re like to help you grow. And when I was with my dad, I can’t remember him doing my injections, but I’m assuming that like he must have. I remember doing my injections round there, and with my dad, if I ever had, like my sugar level dropped, he would call it a case of the floppies, so like my dad would be like oh she’s got the floppies, we’ll get the floppies, we’ll go and get her something to eat quickly, but on that aspect my mum has never, she says when I was a baby I did used to get like low sugar level and
whatnot, but as I got older she was like no, no, she’s growing out of it, she’s growing out of it, and my dad’s always been like no, she still gets like low sugar level and everything like that. But I do sometimes, but a lot of it; it’s not too bad now that I’m older. I can, I don’t have to have like regular snacks or anything like that anymore. It’s generally if I don’t eat, if I forget to have a meal or something and sometimes I just sit there, I can’t move and I feel really sick and I don’t like eating then I have to have, like I can’t just have a drink because I need something a bit more filling, so I have like a milkshake or a yoghurt or something, like a thick yoghurt, and that normally helps, but that doesn’t really happen much now.

INTERVIEWER
Okay, so it as an adult you don’t really get.

MINA_F_25_GH_4’8
No, I’d say that even if I do get them I just work through it now, I just put the hunger pains aside, because I’ve always been a bit funny with my eating anyway, so.

INTERVIEWER
Oh have you?

MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
So you’re a picky eater or?

MINA_F_25_GH_4’8
I wouldn’t say that I’m a picky eater. A lot of it’s to do with like my other medication that I’m on as well, which does alter my eating habits, but I can go through phases that I can like regularly, but I don’t eat breakfast, I can’t eat in the mornings, because I feel sick when I wake up and I just can’t look at food, so I normally, my breakfast is like twelve, one o’clock, which is my lunch. Then I have my lunch normally about three or four o’clock, and then I try to have dinner at five, six or sometimes it can be quite late, but I can quite happily go through the whole morning, go to work, start at like nine, come home, at say four or five o’clock and not have eaten all day, and just had a drink, and I feel fine.

INTERVIEWER
And do you think that’s part of your Russell Silver, like when you’re eating from when you were young or do you think that’s just who you are?

MINA_F_25_GH_4’8
I think that’s just who I am now, but I don’t know, because it’s a bit hard to say, because obviously you don’t, people don’t know with adults with Silver Russell Syndrome, so I don’t know if it is to do with that or if it’s just me or if it’s a bit of both. I’m not a hundred percent sure on that.

INTERVIEWER
No, it’s hard to say isn’t it?
MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
Yeah.

MINA_F_25_GH_4’8
And because I’ve got IBS as well I have to be really careful with what I eat as well. I can’t have nothing spicy, nothing spicy whatsoever, I can’t eat eggs, they like set me off and everything, so I have to be careful with what I eat as well. So it is a bit of a pain, but I’m getting there.

INTERVIEWER
Yeah, it sounds like it. So going back to when you were young, how did Russell Silver affect you when you were young, like when you were at school and things like that?

MINA_F_25_GH_4’8
When I was, like before school, I was fine, I didn’t know much different really, I was just a normal kid, well I had my sister there, I was fine. All I knew is that I was special, and I had this thing, you know I had some special injections to make me grow and I had to eat regularly. But lower school was fine; they were, as far as I was aware, they were really good with that side of things. But because the other, because I’ve got lots of other conditions as well that I didn’t get diagnosed with until I was twenty-one, so I think a lot of that affected me in school as well as the Silver Russell Syndrome, it kind of all added together.

INTERVIEWER
Yes, so how did the other conditions affect you?

MINA_F_25_GH_4’8
Because I got diagnosed with ADHD, so I was constantly very hyperactive, always running about, couldn’t sit still. I’ve got dyspraxia, so my attention span is not always there and especially with the ADHD, trying to focus on something, I’ll just, I couldn’t do it. I have short-term memory loss, so I forget things sometimes, like well I could be watching telly and be like, going on advert, like what are we watching. I’ll just forget what we’re watching. So that was quite difficult, because they just thought I was a problem child, they didn’t know, but it was, apart from that with the Silver Russell side it was alright, in the lower school. Middle school, started getting a of like the bullying, because then I got diagnosed with traits of Asperger’s as well, so that mixed in with the ADHD, and I didn’t, we didn’t know that I had that then, so my head was all over the place, and then I really hated the fact that I was little. I hated in school, because I used to get bullied for it. And I hated, and I still hate the fact now that my feet are so small, I just wear size three feet, so I can’t at least the nice, pretty shoes the nice pretty shoes that all the other girls wear and stuff like that, and it’s annoying when you go into a shop and you have to go into the kids section and you’re like I don’t want trainers that flash up, because I’m twenty-five. But I’m getting there with it now, but back in school I hated it. It was mainly height issue; I hated being small, because I had really big hips as well, so I was just really short, with big hips and big thighs and I just, I hated being small.
INTERVIEWER
So how did it feel to hate your stature and things like that; how did that feel?

MINA_F_25_GH_4’8
Because I’ve always had a bit of a, because I got diagnosed with depression as well, which was like for two different things, that I went through a very low time and I was self-harming, because I physically hated the way that I looked, I hated myself and knew there was nothing that I could do about it to change it, and I couldn’t accept the fact that that was who I am. My mum was like ‘but for people to love you, you need to love yourself’ and everything. And I was one of them people that if somebody had just a nice word to me like oh you look nice today, that would have boosted my confidence, but I never got that. I just got bullied constantly, so that really dragged me down, and I was just like ‘why would anybody like this, why would anybody want to go out with this’, like.

INTERVIEWER
Gosh, that’s what you felt about yourself?

MINA_F_25_GH_4’8
Yeah, and I absolutely hated it, so I would just go out drinking and just get smashed off my face and I put my mum through so much, bless her. She’s been a real saint with me, I’m telling you.

INTERVIEWER
So what kind of age was this happening, where you were really hating the way you looked?

MINA_F_25_GH_4’8
It’s about thirteen, so I was a teenager, right up until I’d say when I was about twenty, so quite a long time that I was feeling like that and then with bad relationships then I started to just feel used and not that anybody liked me for me and I thought well if nobody can like me, then how am I meant to like me for me. And then it was just simple things, like that I can’t wear, most bracelets don’t fit me, I can’t wear bangles, and that really.

INTERVIEWER
Because they just come off.

MINA_F_25_GH_4’8
They just slide down.

INTERVIEWER
Yeah, okay.

MINA_F_25_GH_4’8
And if I’m walking I’ll have to hold my hand out like that so they don’t fall down.

INTERVIEWER
Become loose.
MINA_F_25_GH_4’8
And going out, all the girls, all my friends that are quite tall anyway, well normal height, and they’d put on their high-heeled shoes and we would go out and they’d be like all the way up there, and I’m just down there and constantly getting ID’d as well. That is, even when they look at my passport or whatever and they’re like that’s not you, and I get so defensive, because it’s just because I’m small, so I do think a lot of the time it’s because I’m small I can’t do a lot, like it is difficult to do stuff, it really is.

INTERVIEWER
So it’s stopped you from doing things in the past.

MINA_F_25_GH_4’8
Yeah, yeah. I wouldn’t want to do stuff because I wouldn’t want to go the gym because I know I couldn’t lift my arms up high enough to pull the stuff down and I hated going shopping and everything like that.

INTERVIEWER
Why can’t you?

MINA_F_25_GH_4’8
Because everything’s sort of really high and I’d sit down and be like I can’t reach it, so what I do, and that would just, it was embarrassing to me and then I just wouldn’t want to go out and do stuff. And then I’d find that with my Asperger’s traits and everything, because it took like, because I wouldn’t, I’m not dumb, but me it takes me a while to get stuff. I’ll get there eventually, and because of that I was hanging out with kids that were like younger than me, so then when I had caught up to what my actual age was nobody wanted to know me because they were like she, they associated me, she’s the girl that hangs around like the kids, so she’s a kid type of thing. So that was really difficult as well, but and that I didn’t really go to other school. I just skipped it a lot. I did struggle tremendously in upper school. I hated school.

INTERVIEWER
So what were the main reasons that you struggled and hated school, what was it really?

MINA_F_25_GH_4’8
Mainly for the height again, because I used to still get bullied there. And it was down to the, I should have had a lot more support in school than I had and they, my psychologist said that if I’d been diagnosed earlier with everything I would have done a lot better in life, and it was mainly doing the maths side of things and sports. Because of the Silver Russell Syndrome I would get tired more easily than other kids, so doing running I couldn’t keep up with them. When I was little, like I couldn’t push the Hoover across the floor because it was too hard to do, and so while everybody else is out doing stuff and I’m going to bed early because I’m absolutely shattered from the days. Going to sleepovers what always a nightmare, because I’d fall asleep about nine, ten o’clock and everybody else was still awake and everything. And then with the Silver Russell Syndrome I can’t eat late. If I eat pasta, if it was about nine o’clock I can guarantee that I will throw up, and I still do that occasionally now, I used to do it all the time when I was little. So again going to sleepovers was really difficult, because there’d be the risk of if you’re in a bunk-bed Mina has to sleep on the bottom, she can’t go on the top because in case she has to go to
the bathroom, and if people stayed I had to have like, you know the bathroom steps, like
when you’re little, like the little tub things, we had one of those and I would call it my sick
bucket and then I’d just turn it over and be sick and so that mum would come and clean
it, but then that was always on my bed, in case I needed it, so having people coming
overnight, oh what’s that. And I remember when I stayed over at my friends house and
about ten o’clock at night, well about two in the morning I just, and it was on a bunk bed,
I just threw up everywhere, we were sleeping top to tail, and she had to wake her mum
up, had to change the bed sheets and it was just embarrassing, more than anything that I
couldn’t just do simple things like other kids could, which was difficult.

INTERVIEWER
And would that stop you then going on sleepovers and things like that?

MINA_F_25_GH_4’8
Yeah, I wouldn’t want it, I’d, people would invite me and I remember then, because I’d
feel bad about it, but it got to the point that ‘cause I’m not very good at telling people no,
I feel really, I don’t want to hurt them, so it got to the point that I’d say to my mum, can
you pretend that you’ve grounded me and that I’m not allowed to go out. So she’s come
in and be like [name] you can’t go out, you’re grounded because you’ve done this, when I
hadn’t done any of it, I just didn’t want to go out. So it got to the point where I was lying
to my friends as well just because I was too embarrassed in case something happened
basically.

INTERVIEWER
Yes, oh gosh. And so when you said you were like bullied at school, what kind of things
would people say to you?

MINA_F_25_GH_4’8
Just, I can’t really remember a lot of it, but a lot of it was mainly about my height, like oh
you’re so small, why are you so small, you’re tiny, why can’t you can’t grow; everything
like that. And it was just really upsetting ‘cause I wanted, because even if you’d say to
them oh I’ve got a medical condition, what I found is because nobody’s really heard of
Silver Russell Syndrome half of the people won’t believe you and half of the people don’t
have the common decency to turn around and say what’s that, can you explain to me,
and at lot of, because if you say oh it’s Down’s syndrome everybody knows what that is,
so you’re like oh okay, there’s a bit more understanding, but because they don’t know
what it is and then that makes you seem even more strange and more weird to them,
that you’ve got something and that’s why you’re small, and they don’t, and that’s what,
but that’s just what school kids are like though. They don’t turn, especially all the boys,
they’re not going to try and be like what’s that about, talk to me, you know, they’re not
going to do that all, no.

INTERVIEWER
No, not mature enough.

MINA_F_25_GH_4’8
So that was really difficult. And I found because I was like a bit slow in school as well, and
my sister’s so clever, she is seriously smart, like I’d just, I skipped the generation on that
gene I’m telling you, like all A’s, A-stars, through out school, and there’s me like with
struggling to get a C with my D’s and E’s and F’s, so I found that really difficult. Because when I got to school, everybody was like oh yeah, [name] little sister, oh she’s going to be clever like her sister, and when I wasn’t, they were a bit like oh, what’s the matter with you, why aren’t you clever like your sister. And that was really difficult as well.

INTERVIEWER
So you felt like you were compared to your sister?

MINA_F_25_GH_4’8
Yeah, and I was, and like when I was, all I wanted to do was be a be like my sister, but I still do, like she’s my world, she really is, she’s my big sister and she’s always been there for me. We’ve had our arguments, had our fights, but she’s always been there for me and I know she always will be. And when I was, because I idolised her so much I wanted to be like her and that upset me when I couldn’t do things. Because she’s so talented at drawing, at doing art, anything arty, and I can’t even draw a stick figure straight, so like now we laugh about it, I’m just I can’t draw, but back then it really upset, I was like why can’t I do something good like she’s doing. And how she’d like make friends so easily and she was bubbly and that got to me, like why can’t I make friends as easy as she can, why am I struggling where she’s not. And then it was my mum that turned around and said she’s not as lucky as you think, she has her problems with her friends and everything. And that kind of, it didn’t burst my bubble, but it made me realise that she’s human, if that makes sense, that she’s vulnerable like everybody else, then I got protective over her. So now we just look after each other, but now I just admire her for what she does. I’m not jealous of her, I wouldn’t want to do what she does, she’s too busy; I’m far too lazy for that, but I admire her, because she’s just, she’s just so smart, she really is, she’s just so talented and she’s just an amazing person, like she really is.

INTERVIEWER
Right, okay.

MINA_F_25_GH_4’8
I skipped the generation; I skipped the gene on that one.

INTERVIEWER
So it sounds like kind of thirteen and upwards was pretty hard for you.

MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
How do you think that’s impacted on your life now, is it not impacted at all or?

MINA_F_25_GH_4’8
It’s made me a better person now that I’m older and now that I’m on my medication, and now that I’ve got a group of friends that I can count on one hand, and before I used to be like I need more than that, but now that’s all I need. And they’re very, my best friend, she is so understanding, she’s been to the Growth Foundation with me a few times, with my goddaughter, her little girl’s my goddaughter, and she is so understanding, and she doesn’t hold me back. Because other people, they’re a bit like of she’s a bit little, like she
can’t do that. And my other friend, [name], I’ve been friends with her for about ten years, and they’re, because she’s, because [name] has got really bad ADHD as well, so she’s very protective over me, because she’s like ‘Mina you’re so little, like you’re not a violent person’, and like if anybody upsets me she’s straight over there, like ‘what are you doing, leave her alone’, but then they push me to do stuff at the same time. They protect me, but they let, they push me to do more than what I can do. And they’ve always said to me, the nicest thing that [name] has ever said to me, she said ‘Mina you’re not dumb, it just, she’s like you’re the same as me, it just takes you a while to understand stuff, and she was like the things you come out with, you’re really clever, but people don’t see that. And the way that I see it, is that, because I used to, as I say because I used to hate myself so much, but the way that I see it now is that yeah I’m different, because my boyfriend says I’m crazy, but I’m good kind of crazy, not a bad kind of crazy, I’ve got my tablets, I’ve got a psychiatrist, I’m fine, but I think I just see things differently from other people. I see the nicer things, I see things other people don’t see, and I like to think of it that way. That gives me a buzz that I’m not like everybody else, I’m different and I’m proud of it now. I, as a person, I’ve finally figured out who I am, as a person, and having like my boyfriend there and my best friend and my little goddaughter and my mum and my sister, that’s all I need; it really is, like that’s enough for me now.

INTERVIEWER
So it seems like you’re really happy with your life as it is now.

MINA_F_25_GH_4’8
Yeah, it is still like a few ups and downs and that, but it’s a lot less drama in my life now, that that helps. But now it’s just working on my stress levels and, because with my ADHD I still get like really angry, so it’s just I’m trying to find coping mechanisms, which I find at the minute, if I just put my headphones in, listen to my music, I’m fine. Just like leave me to chill for five, ten minutes, get my tab out or whatever, and I’m fine, or throw chocolate at me, worst case scenario, I always find that. Apart from that, it’s all good.

INTERVIEWER
And do you mind telling me a bit more about that difficult time, and you said you kind of self-harmed and you drank; can you tell me a little bit more about that?

MINA_F_25_GH_4’8
Yeah. No, it was, because I found that I was the type of person, it sounds really weird, but I hated to be alone, but I hated to be around people because I wanted my own space. And so, that was before I had my tablets as well, so my head, the best way that I could describe it to the psychiatrist, it was like when I had a down day, I don’t have, I either have a really good day or I have a really down day, there is no in between really. And then there is like if I’m in a really good mood, snap my fingers, I could be in the worst mood in the world, because, just because of something really small, but it really gets to me. And I found some, and I said that the best way to describe it, it was like somebody had come into my brain and removed a piece of it, and I didn’t quite know how to function properly, I didn’t know what to think, I didn’t know what to do. Nothing was exciting to me, but, and then I like to do new things and that are exciting, but because of my Asperger’s traits, I love routine. I have to have routine. So it’s a constant battle with myself still, because I’m pushing myself, oh come on yeah let’s do like something new, and then the other part of me like, no, let’s just stick to what you know, it’s fine you
know, which I don’t mind half the time. I have to have routine because otherwise I’ll go into all sorts of panic, if there’s something new. I don’t like change. If there’s something new, I, it all kicks off, but then when I started self-harming I just, that was, it felt good, it felt like it was a whole load of pressure was just released and it felt really good to do it, and then I’d feel really bad after I’d done it and try and hide it. So it really hurt when I went into the shower and people are wouldn’t that pain be enough, but that pain, compared to the pain that was in my head was nothing. The pain in my head was more and I was just trying to do anything to fix that, because I knew it wasn’t right, but nobody believed me. Like my dad has always said that he never, he doesn’t think I’ve got all these problems, he thinks I’m making it up, which he told me that word for word, he’s told my mum that, my sister, I’m just attention-seeking on that front, I’ve got Silver Russell Syndrome but nothing else. So that’s put, I’ve got, it’s a complicated situation with my dad, it really is. Any other normal person would not have nothing to do with him after what, even my mum has said after what he said to you, the things he’s said and what he’s done to you, like she was like any other person would have just gone. But I said he’s my dad, and I hate him but I love him, but it’s because he’s the only dad I’ll ever have, which is difficult, because growing up I was always a daddy’s girl, always, and I never really got on with my mum. We’d have so many arguments and the one day I remembered, because I was sat in my room crying my eyes out because my head just didn’t feel right, couldn’t explain it, and my mum come in and she says oh what’s the matter, and I said my head doesn’t feel right mum, and she was like I believe you, and that was it, that connection then with my mum, the trust that I had with my mum then, since then we’ve been inseparable. I’ll talk to her about anything and everything, like if I’m stressed or whatever, I can ring her up and she’ll be, she’s one of the few people I will actually listen to, and she can calm me down, and if I don’t understand stuff, because she knows me, she knows how to explain it like in Mina terms and whatever, and she like sticks up for me and everything. So then I started to feel a little bit better in myself, but still not quite right, and it was mum that, because I was twenty-one, the doctors were like well she’s a bit too old now, and it wasn’t, and it wasn’t until my mum went in there and said you need to do something now, she is not right. And it was mum pushing that sent me to the psychiatrist, who was like yeah, you’ve got this, this, this, and this, put me on the medication, and then I see a psychiatrist once, I used to him once a month, now I see him once every six months, just for like a check-up, so I’m doing better. So I’m getting there with it, but it’s nice that I know now if I’ve got any problems, any stress, I can just ring like one of my five people. And with my boyfriend, because he’s trying to get me to be a bit more tough, like stick up for myself a bit more, so, but we’ve got a code that if I ever text him saying ‘baby I need you’, he will be, no matter what he is doing, he will come and find me, there and then, because that’s when something really bad has happened. So it’s kind of giving me a bit more independence at the same time, if that makes sense.

INTERVIEWER
Yeah, you know you’ve got that person to rely on.

MINA_F_25_GH_4’8
Yeah, and I don’t, and I haven’t, since like my teenager years I self-harmed a couple of years ago, which that left like a massive, I’ve got a scar on my arm there from it, and that’s kind of a reminder to myself, if that makes sense that you’re not in that place, you don’t need to be in that place, you’ve got no reason to be in that place. And now, I’ll sit
there and I start to get worried and everything, I’m just like Mina, your head does this to you, you need to go and talk to mum, get all the facts first before you freak out and start accusing people of stuff, and if you’re right then you go crazy at that person, but if not, you need to stay calm, don’t over-think it, which that’s, my latest thing is telling myself that it’s your head, it does that to you, it plays tricks on you, you’re better than that. So that, but yeah it’s been, I’m getting there. So I don’t feel the need that I need, and I know that if, like a couple of weeks ago I felt the lowest that I’d felt in a very, very long time, just because so much drama had happened, and I didn’t feel the need to self-harm. Which that was a really big, like mini-achievement in my own head there, and I was like you’ve got this, just have a cry, have some ice-cream, put a film on, cuddle [name], you’ll be fine, cuddle the cat, right, it’s all good, because, well because my cat, it sounds really weird, because she’s just a cat, but when I was going through a really down time and I didn’t leave my flat for like two weeks, and she stayed with me. Every day she sat with me, didn’t leave my side, even now she follows me everywhere, she’s like, she’s like a friend and she doesn’t even, whenever I’m upset she’ll come and sit with me, and that calms me down as well, having animals, and especially cats, I love cats, but it just clams me right down; it’s really good. Or I’ll go and see my goddaughter, because I can’t be upset when she’s there, I just can’t, it’s physically impossible. Because she’ll be like why are you crying, are you okay, and then she’ll go and get you some food, and she won’t let you go, she’ll be like do you want a drink; I’m like you can’t make a drink you’re like three, but thank you all the same, and she’ll go and get her favourite teddy and be like, cuddle it you’ll feel better. And she’s a little cutie-pie, I just can’t be upset when she’s there. She’s too funny, she really is, she’s a little, oh she’s a little pain in the backside, but I love her to pieces, I would not change her for the world, I really wouldn’t.

INTERVIEWER
So it sounds like you’ve got some really positive coping mechanisms now.

MINA_F_25_GH_4'8
Yeah. I still have my occasional meltdown, and then that’s when my boyfriend’s just pulls the duvet back over his head, well I’ll just go back to sleep, leave her to it, but it’s nice because I’ve had chats with like with [name] my partner, and I said to him that, because I do get nervous, if I get into a new relationship, because they, say that they accept the fact that I’ve got all these things wrong with me and I’m sitting there like you don’t and you don’t understand the full affect of how I can get when I wake up in the middle of the night because I’m going to be sick and I start hyperventilating, my breathing will go, [breathes heavily], and that’s even before I’ve woken up, and stuff like that, and I’m just thinking any normal person would run a mile, but you’re going out with a girl that’s got mental health issues and the Silver Russell Syndrome and so much going on, and I’ll get scared that that puts people off me, because I know that my personality, I think I’m like Marmite, you either love me or you hate me, and I can get a bit over the top at times. I don’t mean to, I just get carried away and unless people know that about me, they’re just like well she’s a bit weird isn’t she, so I do pre-warn people when I meet them like if I get too much, tell me to shut up, I won’t take it personally, I know what I get like, but.

INTERVIEWER
Do you think like you’re autism traits and your dyspraxia and your ADHD are related to your Russell Silver?
MINA_F_25_GH_4’8
See I don’t know, because mum said that they’ve started doing, they’ve started doing like research into that, because she said most people that have got Silver Russell Syndrome have got other problems as well, so we don’t, I would really like to know if that is linked to the Silver Russell Syndrome somehow. I would really like to know that, because before mum said anything, I just thought I just had all these problems, but if it’s all linked into one, because I just always thought the Silver Russell Syndrome was just my height and my blood sugar. That’s all I ever thought, because that’s all I got told, and now I’m finding out well you could have like your ADHD and all of this, because it’s linked to the Silver Russell Syndrome.

INTERVIEWER
Do you think that would make a difference to know that, if you knew it was all Russell Silver would that make a difference to you?

MINA_F_25_GH_4’8
It would to me, on a personal level it would kind of fit the pieces back in the jigsaw for me, and it would give me more of an answer and I think that would help me to cope, because then I can do research onto that and find out more stuff, and then I can let other people know as well. And it’s hard to say to people, they’re like why have you got, why do you do that and why do you this and sometimes I don’t know, and it’s hard to explain to somebody if you don’t know yourself. And a lot of the time when I’m upset as well, and [boyfriend] is like why are you upset, and I’m like I don’t know, and he’ll say well what do you mean you don’t know, and I’m like I can’t explain it myself, so am I expecting him to understand it as well. I’m trying to get him to read up about all my conditions and everything, but if I don’t fully understand it how can I expect him to. I can’t, it’s not fair. So I need more research on my behalf for me to then to share with other people and I think then everybody else would start seeing me like mum does, and that would be really helpful.

INTERVIEWER
How do you think your mum sees you?

MINA_F_25_GH_4’8
I don’t know, she just, I don’t know, she just sees me, she sees me for me, and I know that’s really hard to explain, but she sees me for me and I don’t know else to explain it, I don’t. It’s just she just knows me, she just, she knows what I like, she knows how I act and she doesn’t make excuses for it, she’s just like yeah she’s like that because of this, so deal with it, you know. And that’s like really, that’s why I think me and my mum get on so well, as well, because I can go to her for anything. And I said to her that you’re not allowed to the huh face at me, because otherwise I’m gone, and she hasn’t done it yet, so it’s alright.

INTERVIEWER
Could you, I’ve asked this of everyone, because it seems like a key thing, about intimate relationships, or like romantic relationships.

MINA_F_25_GH_4’8
Yeah.
INTERVIEWER
How do you think your Russell Silver has affected that kind of side of things for you?

MINA_F_25_GH_4’8
It hasn’t.

INTERVIEWER
Hasn’t at all?

MINA_F_25_GH_4’8
I’d say if anything, not I’m sorry to say, but guys love it when you’re small. They do, because they’re just like ‘Yeah, I can just throw you around’, I’m like ‘Alright! Okay! Calm down!’ but no, I would personally I would say I’ve never had an issue with it.

INTERVIEWER
No, okay.

MINA_F_25_GH_4’8
Never.

INTERVIEWER
That’s really good. So like you know when all your friends were starting relationships and stuff you were doing the same things as them?

MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
Yeah, okay.

MINA_F_25_GH_4’8
That side of things never had a problem with it.

INTERVIEWER
That’s really good. No, that’s really good to hear. Okay, brilliant! Oh, and what about, like thinking into the future, like having children, starting a family?

MINA_F_25_GH_4’8
Well, well here we go. ‘cause I kept getting, like it was years ago, I kept getting really bad chronic pain, right here, and they were just like oh you just get really bad period pains. And I got to point that I couldn’t get out of bed, I had a heat-pack on, an ice-pack on, Ibuprofen and Paracetamol, and I couldn’t move, and I was like that’s not normal, I can’t move, this is affecting my life, I can’t go to work or whatnot. So they sent me to the hospital, and they said, because to begin with they were like oh you might be having an ectopic pregnancy, so I thought oh! They sent me to the hospital, and she was like, oh you’ve got polycystic ovaries. And that really got to me, because one of the reasons I got diagnosed with depression is because they said with the Silver Russell Syndrome it’s going to be harder to conceive anyway, and then they said with the polycystic ovaries on top of that it’s going to be damn near impossible for you to get pregnant, and that hit me,
because ever since, even when I was in school, like lower school and they were like what
do you want to be when you’re older, I was like I want to be a mum. That’s all I’ve ever
wanted to be, and it got to the point I got so depressed about it, I couldn’t watch babies
on TV, I’d have to turn the adverts over, I got so down about it, it was unbelievable. So
when my best friend was like do you want to be godmother, done! And I’ve been there
like every day of that little girl’s life. I love it, and even [name] is like, she is kind of like
your daughter, and even she now says like you’re like my second mummy and everything,
and which I love. So I went through all that time thinking that I couldn’t get pregnant,
and I finally come to terms with it, I’ve come to peace with it, because I was like there’s
plenty of options out there, and if worse comes to worse, I’ll buy a little puppy, and you
can just mother that, it’ll be fine. And then I kept getting really bad pains still, and then,
so they sent me back to the hospital and had like twenty billion internal scans done,
which is like the most undignifying thing in the world, and then they were like oh you
haven’t got polycystic ovaries, and it was all on my medical notes and everything, from
what this nurse had told me, and she says I don’t know why she told you that, she’s
wrong, she said basically when you’re on your period you developed cysts, and when they
burst, which apparently is the same pain as having appendicitis, she goes like that’s why it
really hurts. So I was like okay, and she says like but so you could get pregnant as well.
So I was like okay, this is good news. And then I was with my ex-partner and then long
story short, he left me and then that was the beginning of October, then two months
later I found out I was pregnant, but I didn’t know who the father was. Complete and
utter mess, because literally the build up to that was when I was still with my ex-partner
[name], and a big cup of coffee like that, was making it for him, literally that big, and I put
water in and I stirred it and the cup literally cracked in three places, and it just went all
over my belly, burnt my belly, to like the first layer of skin, so now I’ve got a scar on my
belly from that, and then literally about a week later my little kitten, Muffin, got run over
and killed, and then I found out I was pregnant, and then Christmas Eve I had a
miscarriage. So I was in such a low place at that point, but then my manager said to me,
but Mina you can get pregnant. And I was like, you’re right, I can get blumming pregnant,
I can do this, so that lifted my spirits up a bit. And then, but it was really cute, because I
was with my goddaughter, who’s saying Auntie Mina has got a baby in her belly, and
when I lost the baby, and we obviously didn’t say anything to her, like if we don’t say
anything she’ll just forget it you know, and I was babysitting her and she just sat next
down to her on the sofa and she just looked at me and she put her hand on my belly, she
went, baby’s gone now isn’t it? I was so, how do you know that like, but having my
goddaughter there, because when I went, had the miscarriage, I had to go down to my
dad’s unfortunately, I just wanted to stay with mum, but I had to go down to my dad’s, so
when I come back I said please take me to [name] house, I need to see [name], I need to
see her, I need to hug her, I need to hug one of my little people, and that made me feel a
lot better, so I’m coming to terms with it now. So I’m coping with it a lot more better like
than I used to. And I know that, because my other friend [name], she’s got three kids,
one is three months, [name] is two next month, and [name] has just turned four, and I
know that if I ever feel like I have a broody moment, she’s just like have them, just take
them, just have them for the night, so I get my little fix of like you know my maternal
instinct as it were, so that does help as well. So, but I think if I didn’t have them, didn’t
have my little people I don’t know what I’d do. Because they’re just the world to me,
they really are. I’ve got about nearly nine adopted nieces and nephews, one real nephew,
and about nine adopted ones and I love them to pieces, I do. I spoil them, I don’t care, I
spend money, I don’t care, they make me so happy, and if it makes me happy I’m like
stuff it, why not. I deserve to happy once in a while as well, so. But it’s good, because my partner does want kids, so that’s reassurance for me as well that he wants to do it properly, he wants to get a house together, and then get a dog and all of that, and I was like that’s actually really sweet, you want to do it proper, and I was well that’s fair enough, so I can wait for that, so that’s not too bad.

INTERVIEWER
Oh so you’ve got that in your future.

MINA_F_25_GH_4‘8
Yeah, that’s how I’m, that’s how I see it, that I do want to have kids, I will have kids. I will, because I’ve wanted kids for so long and God help me, it’s my turn next, so help me God it is.

INTERVIEWER
It sounds like you’ve had a lot of practice with your nine adopted nieces and nephews.

MINA_F_25_GH_4‘8
Yeah, oh I just, oh I love them. It’s just newborn babies though, I don’t like them. It’s their soft spot on the top of the head, what is that. Like [name] it’s proper pulsing, I was like put a hat on or something, and they’re just too fragile, and they’re so precious and because I’m quite clumsy, I don’t want nothing bad to happen. Well [name] is like third kid now, she’s just like, but she’ll be like hold them, hold them, change them, you can do it, go and make their bottle, you can do it, so I’ve made a deal with [name] that if I ever have kids, she’s going to have the newborn baby until it’s about three months and I’ll have [name] and then we’ll do a swap.

INTERVIEWER
That’s a good idea!

MINA_F_25_GH_4‘8
Well I said other than that, you’re moving in. Otherwise my baby will just be in a nappy and naked all the time. Won’t go out anywhere, it’s too dangerous.

INTERVIEWER
That sounds sensible.

MINA_F_25_GH_4‘8
But no, that’s what I plan for my future anyway.

INTERVIEWER
Okay. That sounds awesome. Tell me about work, what’s going on for you like for work and what have you done at work?

MINA_F_25_GH_4‘8
Work at the minute, well my first job, what was my first job, I worked in Toys-R-Us, which was alright, because I was just on the tills, so that was alright, but again, because I wasn’t diagnosed at that point I would get, if somebody was rude, I hate rude people, I can’t stand them, so if somebody got rude with me I’d get really upset about it, and they were
like [name] why are you being like that, you’re being really weird, rah, rah, rah, so then I didn’t want to go back there. But apart from that, it was a good like first job. Then I went to MacDonald’s, and I worked there for about nearly three years, was training to be assistant manager and everything. And I liked it there, but then I used to get away with blue murder in that place, I really did, and then I went and then we call it Mina went off the rails, Mina went a bit crackers and ended up with going to Milton Keynes with some Polish lad, like to go and live with him, just as you do, and then I come back and then I worked in a nursery, which I had some really, really, really, it was the worst job I’ve ever had and I will swear that the lady is the devil incarnate. I just didn’t, when I walk into a room I can instantly tell if there’s an atmosphere, you can feel it in you, and as soon as I walked in there I felt it and I never really got on with people. The lady who owned it, like she’d, she’d never give me my fair do, because again because I was undiagnosed and because of my ADH, I would sit there and stare, just daydreaming, and she was like you’re on all these drugs, oh you’re a danger to the children. I was like, well if I’m a danger to the children.

INTERVIEWER
She thought you were on drugs?

MINA_F_25_GH_4'8
Yeah, because I was sitting there staring into space and it was just because of my ADHD. I was just daydreaming you know, and I said well if I’m such a danger to your children why do you let me do nappy changes and everything if I’m so dangerous. And she fired me a week before Christmas, in the middle of the day, so I’d spent most of the night up making food for like our Christmas lunch in the nursery, didn’t get to stay for that, and then because it was midday, so like all the morning kids were going and the afternoon kids were coming in, so I’m trying to hold back the tears and all the kids are like are you going to come play, you going to come play, and I was like I’ll be there in a minute, and then that made me feel even worse. And then I left there, but it was like one time they had, say it was like that size, that unit, and it had like a padlock thing and you lift it up and it’s just like filled with toys, and I lifted it up, the lock bit, and it come off in my hand, so I’m like oh my God, honestly like I didn’t do that on purpose, well look at the size of me, like come on, and she accused me of breaking it, she thought I’d done it on purpose. I was like look at the size of me, like I can’t physically do that, like are you mad. And then her husband come to fix it and was like you owe her an apology, like you owe Mina an apology because that was broken beforehand, and she was like sorry. And it’s like really, I swear you’re meant to be more mature an adult than me. And I seen her daughter a few months later in Sainsbury’s and she just like gave me a dirty look, and I was just like really, there’s just no need for that is there, so I was quite glad when I left there. Because the room that I worked in, it was lovely, it was fine, but then because it was, the lady who owned it, her daughter, her daughter-in-law and her husband worked there, so it was very much all in the family, and even when she was like oh well if you’ve got any problems with them, even if they’re my family, you can come and talk to me, blatantly not, oh you’re clearly going to take their side against the new girl, so that, I hated that job. And I just didn’t agree with the policies, of how they treated the children, and I just never felt like I belonged there. I always felt really awkward when I was there, like I just wanted to be somewhere else and stuff like that. But the job that I’m in now, it’s the best job I’ve ever been in.
INTERVIEWER
And what do you do now?

MINA_F_25_GH_4’8
Hands down, I love it. I work in a shoe shop/podiatry clinic. I just work in the shoe shop bit, not in the clinic, but I love it. I’ve been there three years now. And it’s the best job I’ve ever had, it is. Because my manager, she’s the best manager, and I love her to pieces, [name], because at this point, I’ve been diagnosed, I’m on my medication and so I’m a bit more calmer anyway, and because I’ve been diagnosed, when I went into the interview, I could say I’ve got this, I’ve got this, I’ve got this, I have got learning difficulties, and then because I thought well there’s no point lying, because if I end up having like one of my little hissy fits and they’re like what is the matter with you and I’m like oh I’ve got this and they’re like, you could have said something. And then at this point I had, I’ve got an Employment and Disability worker as well, so she comes in to work to make sure that I’m alright, has meetings with [name], but because [name] son’s got learning difficulties as well, she’s very understanding with me, and she’s very patient and she’s very calming, and if I do something wrong she doesn’t get huffy and puffy with me, she will literally explain it in every single way possible that she can to me, and if I still don’t understand it she will keeping trying until I understand something. And she’s always said to me, which I found a really, and it’s something so simple, but it’s such a big relief, but she said to me if you do something wrong or if something breaks, she says like and she said don’t panic, tell [name], because if you don’t tell [name], [name] can’t fix it. And that was just, you know I just felt so calm then like it’s okay if I make a mistake, because she can fix it. And so I’m very, I’m very dependent on her, I like having her there. When she goes on holiday for a week, so I’m just going no, no, where’re you going, and she’s like I’ll be back soon. I’m like no, come back please. But she’s just, I love it there, and it’s nice because as I say even when I worked at MacDonald’s it was, I thought it was quite nice, because in the mornings I used to work in the kitchen and everything’s really high, so they used to get the microwave and put it down on the lower shelf for me, they used to move stuff so it was lower for me so I could do it. So I always found like that was really nice. And then at my job now, I can just about reach the shoes, like on the display and everything, so it’s not too bad, but everybody else that works there, I think [name] is the tallest, everybody else is short. That’s why [name] hired me, she was like ‘I’m so glad I hired her, like somebody smaller than me! I was like, yeah that’s my mission in life is to make short people feel good about themselves! Wow, I thought I was short. But no, it is the best job I’ve ever, ever had. I love it. I would not change it for the world. Because I’m only part-time, but I like that, because I do Thursdays, Fridays and Saturdays, but I do a lot of overtime as well, and because I’ve loved the job so much, as soon as they’re like oh can you do this, I’m like yeah I’ll do it, so I’ve shown that I’m reliable as well, so that’s really, and they make me feel part of the group. I don’t feel left out with them. We go out for like work meals and they’ll involve me in stuff, and I’ve never really had that before. So it’s nice, and I’m just sitting there like wow I wish I could back to like thirteen-year old me, and I’d be like it does get better, you’re fine, man up. But I love it, and that’s my Godsend that job, it really is.

INTERVIEWER
Would you like to work full-time?

MINA_F_25_GH_4’8
I would. I would, but I wouldn’t want to get another job, but I would like to do like a few more hours there, but then it’s quite nice because then the Sunday is, because normally when [name] is working, because he’s just had lung surgery done, so I’ve had all that blumming drama, so he’s been off for like six weeks, so I said to him like when you go back to work it’s going to kill me, because I’m used to having you here now, but it’s annoying when he’s working, like Sunday is our day together, we don’t see nobody else, we might get round to my sister’s for a roast or something, but, or we might pop down to [name] but we don’t really do a lot, it’s our cleaning day and whatnot. And then Monday, Tuesday and Wednesday, that’s my days to go socialise, go see people, go out, so I like that, but I think that’s because I’ve got into a routine with it, so. But I probably would like to work more and get a bit more money, but.

INTERVIEWER
So if they offered you full-time?

MINA_F_25_GH_4’8
I would take it. I would, because I know that I can do it, and I know I can get there and it’s fine, so I’d be alright with doing that I think. Because I run the shop by myself sometimes, like I’ll close up or I’ll open up, and like I’ll cash up the till, put the money in the safe, do the banking, do all of that and I’m getting tough with customers now as well. Because it’s just difficult, because they just, some of them are just so rude, and I’ve always been taught by mum, if somebody’s nice to you you’re nice back to them, if somebody’s rude to you you’re rude back to them. You treat people how you are treated. And I also, because I like my job, I can’t tell them, I can’t tell them about themselves, if I didn’t appreciate my job so much I would blumming dash this shoe and your head, I’m telling you, just to make you shut up, but that does annoy me and but then I’ll my mum, mum this lady, oh okay, she’s like what happened like, and I have a vent about it and I feel better you know but oh I just can’t stand rude people, I can’t.

INTERVIEWER
Yes. What difference do you think it would have made if you were diagnosed of all these conditions like when you were really young?

MINA_F_25_GH_4’8
I would have done better at school, I would have had the support that I needed, especially in maths. I would have done a lot better and because, I was about twelve by the time I learnt my two times tables, because I just, and I still struggle with the time now. And it’s only really like the last year that I had my first proper watch, because my mum was well there’s no point getting her one when she was little, she couldn’t tell the time, so, but I still get a bit confused with it, but I’m getting there with it now, so it’s not too bad. But I think I would have done a lot better because I think I would have like to think how I think now, and it’s just knowing what you’ve got and you can put a name to it and you can then say okay I’ve got this, these are the symptoms, these are the conditions, let’s work with that. But before that, you don’t know why you’re acting like that and all you know is that you’re acting strange to all the other children, and children at that age, anybody acts weird, don’t go near them, they’re like the weird ones, you know. And I’m quite an emotional person anyway, I’m a softie, I don’t really get, like I’m not a violent person, I’m not. I might throw plates and whatnot, but I’m not a violent person. I won’t talk about it, because I’m scared about getting hit, I’m just like no, it’s a bit painful, but
you know that’s why I have [name], she sticks up for me bless her. But I think I would have done a lot better in life, to be honest, if I’d been diagnosed earlier, I really do. I really do. I think I would have done better at my GCSEs as well. Because I thought I did really well at them, but I only got like Ds and Es and Fs, so I thought there was no bloody point in me turning up was there really. But I went back, when I’d just started at chiropody, so three years ago, just before I started, I went back and did an Adult Maths and English course, so I’ve not got a C grade in both of those, so I went back and did them, because I thought it’s going to look better on my CV to have some Cs in there at least. So I did that.

INTERVIEWER
Well it’s something you know that you can do that.

MINA_F_25_GH_4’8
Yeah, so I was really proud of myself. Because I went to college as well for a bit, but I just, oh I just didn’t get on with the college. I just didn’t agree with their, how they did things and it just, it, because we had like, when they first started they didn’t have a lot of people doing the animal care course, and when I went back to do like the cloakroom is only about that big, and you only had two rows of pegs, and by the time I went, when I went, they’d had about thirty percent more people joined, so you’d hang all your stuff up, people just come in, take off or trampled on it, and I was like I can’t afford to go out and buy a new bag, a new ring-binder, new stuff every week because people are being rude and just can’t put their stuff neatly on the floor, they’ve got to trample on everything. And I was just like no, I’m not coming to college for that, you know and I didn’t really feel like I was learning much, because of the class that I had was really destructive and they wouldn’t, they’d always be talking over. Sometimes I was like well can you just shut up, I’m just trying to listen to what the teacher’s saying you know, and then they’d be like oh Mina you’re such a geek ra, ra, ra, and I’m just like what do you want me to do, I don’t want to be destructive and get told off, I would like to learn, that’s why I’m here. But no, I just don’t do well in the school environment.

INTERVIEWER
No. But you coped well when you re-took your GCSEs, I guess, was that like Adult Learning or something?

MINA_F_25_GH_4’8
Yeah, so that was literally like in the town centre. So I went there every Monday for like six weeks I think. Because I did like my maths and my English, I did like three weeks of each, and took an exam at the end of it. And she was like Mina you’ve just got a C on your maths. And I was like done, don’t want to re-sit, that is fine, I’ve got a C, I’ll take that, I’m happy with that, so that was all good.

INTERVIEWER
Just talking about like how it affects you, Russell Silver, do you feel like you have to explain it to new people you meet?

MINA_F_25_GH_4’8
Sometimes. Sometimes, because a lot of the time they, it’s always the classic signs of people when I first meet them, it’s oh my God you’re feet are so small, oh my God look at
your hands, they're so tiny. So I'm like 'get it out of your system'. Yeah, every time I meet somebody new. And then it's oh my God you're so small aren't you, oh you're so cute, and I'm like back up a minute, I'm twenty-five, I've got Silver Russell Syndrome, which is a height like disorder basically, and they're like oh, oh okay, but that's just annoying when I'm just like yes, I've got small feet, if I had big feet I'd look ridiculous, like I've got flippers on, and if I had bigger hands I could just swat people, so you know. But other than that, and unless I have like a low sugar level attack and if I'm with [name], she'll just tell people for me. She's very, she's very good at reading me, she knows what I'm thinking before I'm thinking it like well if I go to pieces she's, it's been very good for me. She's toughened me up a bit. But yeah, I just sometimes, it just depends on who you're with really, because most of the time I can get away with oh like is your family small? Yes, we're a small family, then I don't really have to go into it. But sometimes, I will like to make a point to people, especially when they're being ignorant or cocky and stuff, and I'm like well actually let me educate you on something for a moment here, you now and it kinds of puts them in their place. And sometimes it's good to do it, but other times I'm not really fussed, because a lot of people, they're just, nowadays, because if you see that somebody's got something wrong with them, you don't, it's just human nature, you stare at them. And I think a lot of people are trying to get up the courage to say I'm not being rude, but can I ask what's wrong with your child you know. So I think a lot of the time people are too scared to ask that, so I'll just tell them, because it kind of breaks the ice a little bit. And so it's not too bad. Or if there's a completion, like oh I've got this wrong with me, I'm not well now. I've got this, this, this, this and this, top that, and they're like oh yeah, but I'm like, I've got Silver Russell Syndrome, have you even heard of it? They go, no, and I'm unique me, it's all good.

INTERVIEWER
So you use it to your advantage.

MINA_F_25_GH_4'8
Oh yes, I've learnt to. You have to these days.

INTERVIEWER
Quite a lot of the people I've spoken to have had a lot of contact with the healthcare system like from childhood to adulthood. What has your experience been of the healthcare system?

MINA_F_25_GH_4'8
All I remember is like growing up, like going into hospitals, coming out of hospitals, that's just normal to me now, it's just day-to-day, I'm not fussed.

INTERVIEWER
Yeah, you said it doesn't bother you.

MINA_F_25_GH_4'8
I never get, no, I've never been fussed by it, at all, because I'm just used to it. My partner, on the other hand, even the thought of going into hospital, he was like hyperventilating, he was really bad, whereas I'm just like yeah, just get on with it. I'm not fussed by it at all.

INTERVIEWER
No, it doesn’t faze you.

MINA_F_25_GH_4’8
I’m really not. That’s all I’ve ever known, was going to hospital and then once a month or something going down to Great Ormond Street, going on a day trip with my mum on the train to London. Get my little magazine, get my little bag of sweets, and yeah. Never fazed me at all, I don’t mind it. I used to love going to hospitals when I was little; I used to love it, like let’s go, I get to play with the toys, get to watch telly in bed, get food delivered, I was on that; such a lazy child! But no, I’m not fazed at all.

INTERVIEWER
It was a positive experience for you?

MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
Yeah, okay, good.

MINA_F_25_GH_4’8
I’ve never had anything really bad happen in hospital, in all fairness, to put me off. Whereas my partner has, bless him, but I’ve been lucky not to have had a bad experience. I just know that if it’s something to do with my Silver Russell Syndrome I’ve got to go to it, so I just get on with it; they’ve just got to do it. And because I think because it’s from such a young age, when you’re such a young age you’re not fussed by things, you’re not really scared, you’ve got no fear, and it’s when you get older that you start to develop these fears, so I think going from a really young age it’s really helped, because I know, I don’t know any different. And now when other people go into hospital, I’m like what are you doing, well it’s me that goes to hospital, why are you here, like well this is my territory you know, but no, I quite like going to hospitals. Not nowadays, because it’s just not the same, you have to sleep in a ward and there’s people snoring, and I’m just like you’re meant to relax and you’re like, oh, oh, I have to sleep with my headphones I do, in hospital. Well luckily I don’t have to go that much anymore now for the Silver Russell anyway, which I think it’s mainly because I’m an adult. And once you turn into your teens with it, that’s it, you’re kind of dismissed because you’re not a child anymore, so you don’t need as much supervision as it were, but I think you do, you should always, because they don’t know, because of that, because of their not doing that, they we’re now doing, because they don’t know.

INTERVIEWER
Yeah, exactly.

MINA_F_25_GH_4’8
Which is just really frustrating, and why didn’t you think to do this earlier, but then I think but it’s quite a new thing with all these people and to have all the different things so maybe it’s they’ve just been too busy or because it’s a new thing and now they’re waiting until now because they’ve never really done it before. So it’s just, but that’s why I’ve been really excited to do it, because I just, I like talking about it, to like for people to just get experience on it.
INTERVIEWER
Yeah, exactly.

MINA_F_25_GH_4’8
Because I’ve been through it all, me, like.

INTERVIEWER
Yeah, it sounds like it.

MINA_F_25_GH_4’8
Blumming too much!

INTERVIEWER
Can we quickly just go back to your growth hormone?

MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
Do you remember like having any side-effects or how it like impacted on your life having growth hormone?

MINA_F_25_GH_4’8
No, there was no side-effects from it. No. If anything, it was just remembering to take it places if we stayed overnight and then remembering to take it back out the fridge in the morning and I’d have to take it on the school trips, which I didn’t really mind, because I’d go out to the, before bedtime and just go and do it. The only down side was because you had to do it in your leg, and nobody told me you could do it in your belly, so I’d be sitting there like with it, in the teachers staff room, because that’s where the fridge is, and I’m just like I don’t really want to pull down my trousers when there’s a male teacher in the room, so I just used to do it through my jeans.

INTERVIEWER
Oh!

MINA_F_25_GH_4’8
And they were like Mina you can’t do that, you can do it in your belly. I was like well why didn’t you tell me this earlier, it could have saved a whole load of kerfuffle, but apart from that, it was always a good ice-breaker. Now when I’m older and I think about it, it was a good way to start a conversation with someone, and they were like oh what’s that, and it’s like oh wow, this is my magic pen that makes me grow and I found that my sister and my three step-brothers growing up, they would use it a lot as a reference in school talks and everything. Like they did a project and they had to use a family member they would, like my brother done one saying that my sister like has this condition, she has to use a special pen to help her grow and everything. So it’s kind of a good thing.

INTERVIEWER
Yeah.
MINA_F_25_GH_4’8
Helping other people I suppose, in some way.

INTERVIEWER
Oh cool. So down to my closing questions now. What aspect of Russell Silver has had the biggest impact on your life?

MINA_F_25_GH_4’8
Height.

INTERVIEWER
Yeah, you said this to me before.

MINA_F_25_GH_4’8
Height. That’s all it is, it’s the height.

INTERVIEWER
So that’s if you could change anything, it would be the height do you think?

MINA_F_25_GH_4’8
Yeah, just a little bit taller.

INTERVIEWER
And what difference would that make you think?

MINA_F_25_GH_4’8
It would just make me feel better, it would mean that I could wear, or that I wouldn’t have to wear, I love wearing jeggings, because it’s the only thing that I can wear, and I wouldn’t have to get mum to roll my trousers and up and tuck them up at the hem, and I’d be able to have slightly bigger feet and wear the nice, pretty shoes, you know, because they’ve started doing nice kids ones like that look like adult ones now, so if I see them and they fit, I don’t care, I’ll get them there.

INTERVIEWER
Okay, cool you’ve answered that. So what are you hopes and worries for the future?

MINA_F_25_GH_4’8
My hopes, it’s like to have children, to have that perfect family life as it were, but maybe not so perfect; perfect gets boring, and I can’t be dealing with perfect.

INTERVIEWER
There’s no such thing!

MINA_F_25_GH_4’8
Especially not with me about, I’m far too clumsy! But my worries, I don’t know. I don’t like to think about if I’ve got any worries. I just tend generally, I just take each day as it comes. I can’t, because, well as I said, people with ADHS, it’s very hard to plan and to see into the future and that aspect and to actually think of a plan to stick to it, so I just like to
take each day as it comes because I don’t know what’s going to happen and I don’t want to build myself up, get myself all excited, I’m going to do this and then it doesn’t happen. Because then I don’t want to let myself down, because I hate feeling like that, but, so I’d say that.

INTERVIEWER  
Yeah, to take each day as it comes.

MINA_F_25_GH_4’8  
Each day as it comes, it’s easier.

INTERVIEWER  
People that have participated in this study have given a lot of their time, so you came all the way down to Southampton and you spent three or four hours with Kemi. And now you’re talking to me about you now you told me some kind of personal things. What does it mean to you to take part in this research?

MINA_F_25_GH_4’8  
Everything. So if it’s going to help future generations because we just don’t know anything, and if I lived through it, and if what I’ve been through helps other people, so they don’t maybe feel the way that I feel, they have a bit of closure in their life, then that’s what I would have wanted, so if I could do that for somebody else, then why not. And I don’t, I can’t help with a lot of things, and this is one thing I can help with, so I’m going to do it. Because I’m not exactly good at doing many things, and this is one thing that I can do, and I can do it right, so.

INTERVIEWER  
Yeah, brilliant.

MINA_F_25_GH_4’8  
It doesn’t happen a lot!

INTERVIEWER  
Okay, well that’s all my questions. Is there anything you want to add or is there something that you thought I might ask and I didn’t?

MINA_F_25_GH_4’8  
I don’t think so.

INTERVIEWER  
No?

MINA_F_25_GH_4’8  
No.

INTERVIEWER  
Happy to draw it to a close?

MINA_F_25_GH_4’8
Yeah.

INTERVIEWER
Happy for me to turn the tape-recorder off?

MINA_F_25_GH_4’8
Yeah, yeah, fine.

INTERVIEWER
Awesome.
Appendix 9 – Buddy system for researcher safety

STAARS Qualitative Study Lone Worker Document

The diagram below outlines the procedure for lone working in the STAARS qualitative study and has been informed by the University Hospital Southampton Foundation NHS Trust’s Lone worker policy dated 24th November 2010 and the ‘Not alone: A guide for the better protection of lone workers in the NHS’, 2009. Relevant information from each document has been included below to provide more detail if required. However the lone worker should read both documents in full as these include information on safety responsibilities.

Step 1
• Lone worker (LW) to let buddy know the date, time and location of participant interview in advance to ensure availability. If designated buddy is not available an alternative must be sought.

Step 2
• LW to call buddy before going into interview to advise start of lone working and expected time to finish
• Buddy to set reminder to expect call from LW by stated time

Step 3
• LW to call buddy when lone working finishes. A discreet code word will be used if there is a problem, which is “Hi, Clinical Genetics/ Hi, is that Clinical Genetics/Hi, can you put me through to Clinical Genetics?”
• If the buddy does not receive a call from the LW by the designated time the buddy will contact the LW

Step 4
• If the buddy cannot contact the LW, the buddy will try to contact the participant
• If the buddy cannot contact the LW or the participant, the escalation plan will be implemented (see below)

Escalation Plan:
Line manager - Angela Fenwick (find out which number is best to use)
Service Manager - Prof Karen Temple (07787 651 440)
The Police

What to do if buddy is sick/not available/leaves work before LW finishes:
Buddy designates alternative

Lone worker next of kin:
Husband – Aaron Ballard (07977 409 869/02380 230 194)
Parents – Jill and Peter Hodges (07815 815 289/02380 892 340)

Lone workers vehicle details:
Dark blue Ford Fiesta – ND05 RXT
Lone Worker Policy UHS 24.11.2010

5.6 Lone worker movements
Someone away from the Lone worker should be aware of the lone workers whereabouts, who they are visiting, what is their location and the estimated time scale that the Lone worker will be with the relative or patient. A buddy system may be appropriate for some staff groups. A full escalation plan should also be known to staff who are not with the Lone worker to ensure that in the event of an incident e.g. failure to return or next patients not visited the escalation plan can be activated. Appropriate activation stages should be used, a line manager, senior manager and, ultimately, the police. Any individual nominated as an escalation point should be fully aware of their role and responsibilities. Each department must have their own local activation procedures. Please refer to the Trust Incident Reporting and Management Policy for Future information http://www.nhsbsa.nhs.uk/Documents/SecurityManagement/Lone_Working_Guidance_final.pdf

6. Preparing for lone working
6.15.1 Before visiting a location or patient/service user that is a known risk, colleagues who may have worked alone in the same situation previously should be contacted. This aids communication and informs the action taken to minimise the risks.

6.17.1 Lone workers should always ensure that someone else (a manager or appropriate colleague) is aware of their movements. This means providing them with the address of where they will be working, details of the people they will be working with or visiting, telephone numbers if known and expected arrival and departure times.

6.17.2 Lone workers should leave a written visiting log, containing a diary of visits, with a manager and colleague(s). This information must be kept confidential. Details can be left on a whiteboard or similar, if it is in a secure office to which neither patients/service users nor members of the public have access.

6.17.3 Arrangements should be in place to ensure that if a colleague with whom details have been left leaves work, they will pass the details to another colleague who will check that the lone worker arrives back at their office/base or has safely completed their duties. For office-based staff, if details have been left on a whiteboard, they must not be erased until it has been confirmed that the lone worker has returned safely or completed their duties for that day.

6.17.4 Details of vehicles used by lone workers should also be left with a manager or colleague, for example, registration number, make, model and colour.

6.17.5 Procedures should also be in place to ensure that the lone worker is in regular contact with their manager or relevant colleague, particularly if they are delayed or have to cancel an appointment.

6.17.6 Where there is genuine concern, as a result of a lone worker failing to attend a visit or an arranged meeting within an agreed time, or to make contact as agreed, the manager should use the information provided in the log to locate them and ascertain whether they turned up for previous appointments that day. Depending on the circumstances and whether contact through normal means (mobile phone, pager, etc)
can be made, the manager or colleague should involve the police, if necessary (see escalation process in 6.19).

6.17.7 If it is thought that the lone worker may be at risk, it is important that matters are dealt with quickly, after considering all the available facts. If police involvement is needed, they should be given full access to information held and personnel who may hold it, if that information might help trace the lone worker and provide a fuller assessment of any risks they may be facing.

6.17.8 It is important that contact arrangements, once in place, are adhered to. Many such procedures fail simply because staff forget to make the necessary call when they finish their shift. The result is unnecessary escalation and expense, which undermines the integrity of the process.

6.18 The buddy system
6.18.1 It is essential that lone workers keep in contact with colleagues and ensure that they make another colleague aware of their movements. This can be done by implementing management procedures such as the ‘buddy system’.

6.18.2 To operate the buddy system, an organisation must ensure that a lone worker nominates a buddy. This is a person who is their nominated contact for the period in which they will be working alone. The nominated buddy will:

- be fully aware of the movements of the lone worker
- have all necessary contact details for the lone worker, including next of kin
- have details of the lone worker’s known breaks or rest periods
- attempt to contact the lone worker if they do not contact the buddy as agreed
- follow the agreed local escalation procedures for alerting their senior manager and/or the police if the lone worker cannot be contacted or if they fail to contact their buddy within agreed and reasonable timescales.

6.18.3 The following are essential to the effective operation of the buddy system:

- the buddy must be made aware that they have been nominated and what the procedures and requirement for this role are

- contingency arrangements should be in place for someone else to take over the role of the buddy in case the nominated person is unavailable, for example if the lone working situation extends past the end of the nominated person’s normal working day or shift, if the shift varies, or if the nominated person is away on annual leave or off sick.

6.19 Escalation process
6.19.1 It is important for NHS organisations to have an escalation policy and process, outlining who should be notified if a lone worker cannot be contacted or if they fail to contact the relevant individual within agreed or reasonable timescales. The escalation process should include risk assessment and identification of contact points at appropriate stages, including a line manager, senior manager and, ultimately, the police. Any individual nominated as an escalation point should be fully aware of their role and its responsibilities.
9. Technology
9.4 Practical suggestions on the use of a mobile phone
9.4.7 In some circumstances, agreed ‘code’ words or phrases should be used to help lone workers convey the nature of the threat to their managers or colleagues so that they can provide the appropriate response, such as involving the police. The decision to use code words or phrases should give due consideration to the ability of a member of staff to recall and use them in a highly stressful situation.
Appendix 10 – Letters confirming ethical approval (NRES and UWE)

19 December 2013

Professor I. Karen Temple
Professor of Medical Genetics
University of Southampton and University Hospital Southampton
Wessex Clinical Genetics Service
Princess Anna Hospital, Cowford Road,
Southampton, Hampshire.
SO16 5YA

Dear Professor Temple

Study title: Study of Adults and Adolescents with Russell Silver syndrome in the UK
REC reference: 13/S6/0630
Protocol number: RHM NEU0209
IRAS project ID: 132544

The Research Ethics Committee reviewed the above application at the meeting held on 11 December 2013. Thank you for attending attended with Dr Kemi Lokulo-Sodipe and Dr Justin Davis to discuss the application:

1. The Committee sought clarification that you will be offering a new genetic test for participants in the study.

You stated that you will be performing genetic tests on all participants in the study to confirm that they do have a correct diagnosis of Russell Silver Syndrome (RSS). You stated that many adults with RSS may not have had their diagnosis confirmed by genetic tests as only about 90% of patients with RSS have had a proper genetic diagnosis. You confirmed that some participants in the study may find that they do not have the known genetic cause for their RSS diagnosis. You stated that it is thought that as many as 20% of patients with RSS will have actually been diagnosed in some other way. You confirmed that you will be recruiting participants to the study through clinical genetics.

2. The Committee sought clarification of the possibility of a negative genetic test, and the implications that this would have for the participants.

You clarified that a negative genetic test does not mean that the patient does not have RSS, but only that they do not have this particular genetic cause for their condition.

3. The Committee sought clarification of the main aims of the study.

A Research Ethics Committee established by the Health Research Authority
Health Research Authority

You stated that you have 3 main aims. The first main aim is to try to correlate participants' final heights to whether or not they have received growth hormone treatment. The second is to try to assess the prevalence of any other medical indications, such as diabetes. The third aim is to assess patients' quality of life with RRS.

4. The Committee queried whether you would also be assessing the impact of administering growth hormone therapy by means of daily injections on quality of life.

You confirmed that, as well as looking at height indicators and the effect of growth hormones on height, you will be assessing the impact of the growth hormone therapy on quality of life during the in-depth interviews.

5. The Committee queried whether you were assessing parental height.

You confirmed that 90% of a person's height is determined by genetics from their parents, so you will be assessing participants' parental height.

6. The Committee queried whether the publishing of the medical photographs is optional.

You confirmed that this is optional, and if a participant declines to have their photographs taken or for them to be published, they will still be able to participate in the main study.

The Committee stated that the Consent Form may need clarifying in terms of the items that are optional.

You agreed to this point and stated that making this clear could enhance recruitment.

Ethical opinion

The members of the Committee present gave a favourable ethical opinion of the above research on the basis described in the application form, protocol and supporting documentation, subject to the conditions specified below.

We plan to publish your research summary wording for the above study on the NRES website, together with your contact details, unless you expressly withhold permission to do so. Publication will be no earlier than three months from the date of this favourable opinion letter. Should you wish to provide a substitute contact point, require further information, or wish to withhold permission to publish, please contact the REC Manager, Libby Watson, at mrescommittee.southcentral-hampshire@nhs.net.

Ethical review of research sites

NHS Sites

The favourable opinion applies to all NHS sites taking part in the study, subject to management permission being obtained from the NHS/HSC R&D office prior to the start of the study (see "Conditions of the favourable opinion" below).
Conditions of the favourable opinion

The favourable opinion is subject to the following conditions being met prior to the start of the study:

1. The Committee requests that the Response Form is amended so that it is opt-in only, so that ‘Please delete as appropriate’ and ‘I would not’ are removed, as well as the phrase ‘If you would like to be contacted’ appearing immediately before the request for contact details.

2. The Committee requests that the Reply Slip at the end of Part 1 on page 7 of the Participant and Parent/Guardian Information Booklet is moved to the final page in such a way that it can be separated from the rest of the information booklet for return purposes.

3. The Committee requests that the Contact Details at the end of Part 1 of the PIS for Relatives should be removed, with the email address added to the Contact Details at the end of Part 2.

4. The Committee requests that in both PISs the first sentence of the section ‘What are the possible benefits of taking part?’ is amended with the word ‘will’ changed to ‘may’, and with the last part of the sentence changed to read ‘in the hope of improving knowledge so that appropriate treatment might be offered to people with this condition’.

5. The Committee requests that the Consent Forms are amended as follows:
   a. Any optional items are clearly labelled as such, with ‘yes’ and ‘no’ boxes, and moved to the section at the end of the Consent Forms.
   b. Item 3 on the Consent Form (Item 4 on the In-depth Interview Consent Forms) should be amended to read: ‘I understand that relevant sections of [my/your child’s] medical notes and data collected during the study, may be looked at by individuals from regulatory authorities or from the [name] NHS Trust, where it is relevant to [my/your child’s] taking part in this research. I give permission for these individuals to have access to [my/your child’s] records’.
   c. An item should be added for consent to take the medical photographs for research purposes (which is separate to seeking consent to publish them).
   d. Item 4 of the Consent Form for participating relatives is amended to read: ‘I give consent for the research team to access my medical records if necessary to obtain data relating to this study. I understand that this data will only be used for the purposes of this study, and may be anonymously reported in the study results when they are published (which may be online).’
   e. In each Consent Form when referring to ‘future research’ or ‘ethically approved studies’ this should be qualified by adding ‘related to RSS’.
   f. In the Consent form for Participating Relatives item 5 replaces ‘spit’ by ‘saliva’.

6. The Committee requests that the picture is removed from the top of the Assent Forms as it is deemed to be too juvenile for the age group in question.

7. The Committee recommends that you consider producing separate Adult Participant and Parent/Guardian Information Booklets, so that they can refer correctly to “you” and “your child” throughout.
Health Research Authority

You should notify the REC in writing once all conditions have been met (except for site approvals from host organisations) and provide copies of any revised documentation with updated version numbers. The REC will acknowledge receipt and provide a final list of the approved documentation for the study, which can be made available to host organisations to facilitate their permission for the study. Failure to provide the final versions to the REC may cause delay in obtaining permissions.

Management permission or approval must be obtained from each host organisation prior to the start of the study at the site concerned.

Management permission ("R&D approval") should be sought from all NHS organisations involved in the study in accordance with NHS research governance arrangements.

Guidance on applying for NHS permission for research is available in the Integrated Research Application System or at http://www.rdfforum.nhs.uk.

Where a NHS organisation's role in the study is limited to identifying and referring potential participants to research sites ("participant identification centres"), guidance should be sought from the R&D office on the information it requires to give permission for this activity.

For non-NHS sites, site management permission should be obtained in accordance with the procedures of the relevant host organisation.

Sponsors are not required to notify the Committee of approvals from host organisations.

Registration of Clinical Trials

All clinical trials (defined as the first four categories on the IRAS filter page) must be registered on a publicly accessible database within 6 weeks of recruitment of the first participant (for medical device studies, within the timeline determined by the current registration and publication times).

There is no requirement to separately notify the REC but you should do so at the earliest opportunity e.g. when submitting an amendment. We will audit the registration details as part of the annual progress reporting process.

To ensure transparency in research, we strongly recommend that all research is registered but for non-clinical trials this is not currently mandatory.

If a sponsor wishes to contest the need for registration they should contact Catherine Blewett (catherineblewett@nhs.net), the HRA does not, however, expect exceptions to be made. Guidance on where to register is provided within IRAS.

It is responsibility of the sponsor to ensure that all the conditions are complied with before the start of the study or its initiation at a particular site (as applicable).

Approved documents

The documents reviewed and approved at the meeting were:

A Research Ethics Committee established by the Health Research Authority
<table>
<thead>
<tr>
<th>Document</th>
<th>Version</th>
<th>Date</th>
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<td>Covering Letter</td>
<td></td>
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</tr>
<tr>
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<td>Telephone Prompt Form, v1</td>
<td>08 November 2013</td>
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<td>Prof Temple</td>
<td>19 November 2013</td>
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<td>Letter from Sponsor</td>
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<td>26 September 2013</td>
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<tr>
<td>Letter of invitation to participant</td>
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<td>06 November 2013</td>
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<td>Other: Study Visit Outcome Sheet</td>
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<td>Participant Information Sheet: Adults/Parents and Guardians Information Booklet</td>
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<td>Participant Information Sheet: Young People</td>
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<td>Questionnaire: Sheehan Disability Scale</td>
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<td>Questionnaire: Schedule for the individual evaluation of QOL-direct weighing</td>
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<td>Questionnaire: Male puberty self-assessment questionnaire</td>
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<tr>
<td>Questionnaire: Female puberty self-assessment questionnaire</td>
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<tr>
<td>Questionnaire: Early Medical History Questionnaire for parent of STAARS participant</td>
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<td>REC application</td>
<td>132644</td>
<td>11 December 2013</td>
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<tr>
<td>Reference or other scientific critique report</td>
<td>NIHR Lay and Peer Review Forms</td>
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Membership of the Committee

The members of the Ethics Committee who were present at the meeting are listed on the attached sheet.

Alison Bower declared that she had advised the CI on seeking patient and public involvement in the design of the research. The Committee agreed that this did not present a conflict of interest.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

After ethical review

Reporting requirements

The attached document "After ethical review – guidance for researchers" gives detailed guidance on reporting requirements for studies with a favourable opinion, including:

- Notifying substantial amendments
- Adding new sites and investigators
- Notification of serious breaches of the protocol
- Progress and safety reports
- Notifying the end of the study

The NRES website also provides guidance on these topics, which is updated in the light of changes in reporting requirements or procedures.

Feedback

You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.

Further information is available at National Research Ethics Service website > After Review

13/SC/0630 Please quote this number on all correspondence

We are pleased to welcome researchers and R & D staff at our NRES committee members' training days – see details at http://www.hra.nhs.uk/eire-training/

With the Committee's best wishes for the success of this project.
Yours sincerely

[Signature]

Professor Ron King
Chair

Email: nrescommittee.southcentral.hampshire@nhs.net

Enclosures: List of names and professions of members who were present at the meeting and those who submitted written comments. "After ethical review - guidance for researchers" [SL-AR2]

Copy to: Dr Olukareni Lokuo-Sadjo
Mia Hadi Naldi, University Hospital Southampton NHS Foundation Trust

NRES Committee South Central - Hampshire B

Attendance at Committee meeting on 11 December 2013

Committee Members:

<table>
<thead>
<tr>
<th>Name</th>
<th>Profession</th>
<th>Present</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mrs Ila Berry</td>
<td>Clinical Psychologist (Retired)</td>
<td>Yes</td>
</tr>
<tr>
<td>Mr Brian Birch</td>
<td>Consultant Urological Surgeon</td>
<td>Yes</td>
</tr>
<tr>
<td>Mrs Alison Bowser</td>
<td>Patient and Public Involvement Officer</td>
<td>Yes</td>
</tr>
<tr>
<td>Mrs Janet Brember</td>
<td>Pharmacist</td>
<td>Yes</td>
</tr>
<tr>
<td>Dr Diane Carpenter</td>
<td>Lecturer, Mental Health Studies</td>
<td>Yes</td>
</tr>
<tr>
<td>Mr Mark Cassidy</td>
<td>Lecturer in Radiography</td>
<td>Yes</td>
</tr>
<tr>
<td>Mrs Angela Iveson</td>
<td>Oncology Research Nurse</td>
<td>Yes</td>
</tr>
<tr>
<td>Professor Ron King (Chair)</td>
<td>Mathematician (Retired)</td>
<td>Yes</td>
</tr>
<tr>
<td>Mr Geoff Lowndes</td>
<td>Chartered Engineer (Retired)</td>
<td>Yes</td>
</tr>
<tr>
<td>Dr Chris Markham (Alternate Vice-Chair)</td>
<td>Principal Lecturer - Public Health, Social Care and Research Design Lead</td>
<td>Yes</td>
</tr>
<tr>
<td>Dr Ian McAndrew</td>
<td>Independent EU Consultant/Lecturer</td>
<td>No</td>
</tr>
<tr>
<td>Dr Karl Nunkesing</td>
<td>Principal Psychology Lecturer</td>
<td>No</td>
</tr>
<tr>
<td>Dr Andrew Scott</td>
<td>Course Leader, M.Sc. Clinical Exercise Science</td>
<td>Yes</td>
</tr>
<tr>
<td>Mr Alan Smith</td>
<td>Barrister's Crt (Retired)</td>
<td>Yes</td>
</tr>
<tr>
<td>Dr Giles Tan (Vice-Chair)</td>
<td>Consultant Psychiatrist</td>
<td>No</td>
</tr>
</tbody>
</table>

Also in attendance:

<table>
<thead>
<tr>
<th>Name</th>
<th>Position (or reason for attending)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Miss Libby Watson</td>
<td>HEC Manager</td>
</tr>
</tbody>
</table>
Dear Lisa

Application number: HAS/15/11/043
Application title: Study of Adults and Adolescents with Russell-Silver Syndrome in the UK (STAARS UK)
NHS Application Number: 13/SC/0630

Your NHS Ethics application and approval conditions have been considered by the Faculty Research Ethics Committee on behalf of the University. It has been given ethical approval to proceed with the following conditions:

- You comply with the conditions of the NHS Ethics approval.
- You notify the Faculty Research Ethics Committee of any further correspondence with the NHS Ethics Committee.
- You must notify the Faculty Research Ethics Committee in advance if you wish to make any significant amendments to the original application.
- If you have to terminate your research before completion, please inform the Faculty Research Ethics Committee within 14 days, indicating the reasons.
- Please notify the Faculty Research Ethics Committee if there are any serious events or developments in the research that have an ethical dimension.
- Any changes to the study protocol, which have an ethical dimension, will need to be approved by the Faculty Research Ethics Committee. You should send details of any such amendments to the committee with an explanation of the reason for the proposed changes. Any changes approved by an external research ethics committee must also be communicated to the relevant UWE committee.
- Please note that any information sheets and consent forms should have the UWE logo.

Further guidance is available on the web:
Please note that the University Research Ethics Committee (UREC) is required to monitor and audit the ethical conduct of research involving human participants, data and tissue conducted by academic staff, students and researchers. Your project may be selected for audit from the research projects submitted to and approved by the UREC and its committees.

Please note that your study should not commence at any NHS site until you have obtained final management approval from the R&D department for the relevant NHS care organisation. A copy of the approval letter(s) must be forwarded to Leigh Taylor in line with Research Governance requirements.

We wish you well with your research.

Yours sincerely

Dr Julie Woodley
Chair
Faculty Research Ethics Committee

c.c. Elizabeth Jenkinson
Appendix 11 – Reflection on consent

Description
I had travelled up the night before to prepare to interview a participant the next afternoon. The participant called the office on the morning of the interview to say that her husband had been taken to hospital the night before with internal bleeding. I called the participant to see what she wanted to do. She first told me what had happened and that her husband had been taken to a hospital an hour away from their home. She then asked if I had left from Southampton yet and if I had she would come home at the time we had arranged as she did not want to let me down. I made a snap decision to say that it was fine and that we would arrange to meet another day, even though inside I was thinking ‘I’ve driven all the way up here and stayed in a hotel overnight, just to drive all the way back again’.

Feelings/reaction
It didn’t feel right to tell the participant I was already there. This would have added to her stress. I felt that it would have been guilt that would have motivated her participation at that point and that consent was not completely of her free will.

Evaluation/Analysis
I had recently seen the meme on Facebook around consent for sex and the metaphor of offering someone a cup of tea. This is what triggered these thoughts around consent.

Conclusions (general/specific)
I think I learnt something new about consent and that it is not just simply someone saying ‘yes’ to participating, there are nuances that the empathetic researcher can pick up on to ensure we are getting the best data from willing participants.
Appendix 12 – Reflective chapter

The final push

I have chosen the (Gibbs, 1988) model, which I also used in my professional skills competency, to aid reflecting on the last two months of my doctorate. I have chosen to focus on these months, not only because they are fresh in my mind, but also because they have also been the most challenging to date.

Description

From September 2016 I could no longer use work time to write my thesis, which I was aiming to hand in by the end of November, as I had to concentrate on writing papers. I had a meeting with my supervisors at the end of September to discuss my key findings, after this meeting I felt really ambivalent. On one hand I felt positive and motivated as my supervisors seemed excited by my findings, thought that I was definitely meeting the doctoral descriptors, and they had also seen more substantial chapters and were confident I was almost finished. But on the other hand, I could see how much work I had to do and that the next two months were going to be really intense. All I could think was how am I going to get through to the end?

I made a plan of work for the following two months. This involved working on my thesis for one hour every day, Monday to Friday and six hours on a Saturday and Sunday. However, I had already been working really hard and did not understand why it was taking me so long to write. I did not have trouble thinking about what to write and I had not been procrastinating. I had arranged to hand a final draft to my supervisors in week three, so planned to spend a week on each section (introduction, results, and discussion (my method had been written and checked previously)). However, by the end of week two I was still working on my results section, so had to delay the hand-in date by two weeks.

Feelings

At the start of the two months I felt in despair. The thought of continuing to work this hard for another two months seemed almost intolerable. This was matched by the determination to get this finished, as it had been weighing on my mind for so long. The feeling of knowing I am near the end, but not quite being there yet was unbearable. I could only bear it when I was actually writing my thesis, if I let myself take a step back to take stock of how far I need to go, I almost wanted to give up right there and then. I also felt sceptical about the hour a day of study, as I was unsure if I would achieve anything in this small amount of time.

By the end of week two, I felt better than I did at the start. At the start I felt a sense of dread at the prospect of all this work, but once I was in the midst of it, it actually felt OK, and the hour was really productive. I was worried that my mental health would suffer so I started meditating and tried to fit in exercise where I could. At the end of week four I took a day off and had a really relaxing day, but by the end of the day I had developed a cold and started to feel really ill. By week six I was easily completing the work, but had stopped exercising and meditating.
Evaluation

I had to make a decision between having a work/life balance and getting my thesis completed sooner. I chose to get my thesis completed sooner because the thought of it dragging out for much longer was worse than not having any time to relax. I feel that this was a good decision and an exercise in delayed gratification. The negative side of this decision was that I did not prioritise my mental and physical health. Working for this many days in a row with only one day off are not the actions of a person who understands the negative impact this could have. This means that my actions are incongruent with my beliefs and priorities. I would not advise a friend to do this, so why have I let myself do this?

Analysis

I have realised that I have so much further to go and finishing my doctorate is the beginning, not the end. I have acknowledged that although I have been studying psychology and working in related fields for fourteen years, I am still at the very start of my career as a researcher and a health psychologist. I have got so much to learn and I need to take it one step at a time. These thoughts made me more eager to get my thesis finished and led to a gruelling work schedule.

I think that my constant re-drafting may be an issue and that I should focus on getting it right first time. I spend a lot of time writing, but do not necessarily think I am writing the right thing. In our professional skills session we were advised that if you get ‘writer’s block’ you should just try and write anything, getting started is the barrier. I never have this problem, I can always write. I think this causes me to write in a style that is not academic enough.

Having the rest of the cohort to study with was invaluable, as no one else understands what you are going through. This makes doing a professional doctorate easier in some ways than a PhD, as even though a PhD student will know others doing PhDs, everyone’s PhD topic is different. On the professional doctorate, having five other people that are doing the same assignments as you and struggling at different times, make you feel that you are not alone. And knowing that means a lot.

I wrote a research journal as part of my doctorate and even though it is best practice for a researcher, I wonder who does. I want to take that tool forward to my next piece of work as it helps to write things down, it means that you do not bombard your over worked supervisor with all your thoughts and feelings, you can take some time to sit with them and work out what is going on and how you are going to deal with it. This then enables you to ask for help if you still need it, in a constructive way.

I am very susceptible to leisure sickness, which is a phenomenon whereby a person gets ill at the weekend or whilst on holiday. This happened to me on the one day off I had, where I caught a cold once I became relaxed. This is another motivator for me to just keep going and maintain the intense working schedule, because I fear I will get ill if I stop. One strategy I have learnt over the years is that once the big piece of work is handed in, instead of instantly relaxing, I try to slowly wind down, by maybe keeping busy with
hobbies and pleasant tasks for a week or two more. This allows for a gradual shift back to working five days a week.

Conclusions

In conclusion, writing my thesis and studying to become a health psychologist has been much harder than I thought. But I have learnt so much and my confidence has grown. I will fortunately not have to encounter this specific situation again, but will hopefully be working on similar research projects in the future. At least these projects will be contained (mostly) to the working week and I can use everything I have learnt throughout this process, about research and myself, to make this journey a successful one. I have forgotten who I am and what I do when I am not doing a doctorate and am looking forward to discovering the answers to these questions too.
## Appendix 13 – Dissemination of research

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<td>Teaching Session to Clinical Genetics</td>
<td>Oral presentation</td>
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<tr>
<td>24/10/2015</td>
<td>Child Growth Foundation convention - Warwick</td>
<td>Oral presentation</td>
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<tr>
<td>23/06/2016</td>
<td>Postgraduate Researchers Conference - UWE</td>
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<td>29-30/06/2016</td>
<td>Appearance Matters 7 Conference - London</td>
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<td>26/08/2016</td>
<td>Division of Health Psychology and European Health Psychology Conference - Aberdeen</td>
<td>Poster presentation</td>
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<td>09/2016</td>
<td>Article published in the Journal of Aesthetic Nursing</td>
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<td>01/10/2016</td>
<td>Child Growth Foundation convention - Warwick</td>
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Teaching Session to Clinical Genetics

STAARS: What is it like to live with Russell-Silver Syndrome? A qualitative study.

- Russell-Silver syndrome (RSS) is a rare genetic condition caused by chromosomal problems (Dinarello et al., 2013).
- It is characterized by:
  - Short stature leading to poor postnatal growth and short stature in adulthood
  - Disproportionately large head
  - Triangular facial appearance
  - Body asymmetry
  - Feeding difficulties (Weilbæk, 2011)

STAARS

- Research Question:
  - What is it like to live with RSS?
- Why?
  - Parents often have difficulties in understanding the syndrome
  - Research shows that there are symptoms, but very rarely describes the impact of these symptoms on the patient (Turnis, 2016)
  - Lack of information in adulthood

STAARS recruitment

- Methods:
  - Semi-structured in-depth interviews
  - Participants: aged 15 and over

Hight Isn’t the number one issue

- He thought that his height wasn’t the number one issue
- He felt that it was the least of his problems
- SSA: Stunting due to slow growth

Masculinity/body image

- M_01_QUOTLE_10: "You know what I mean, I was thinking the other day about being a man, knowing what the role of a man is and thinking that..."
- M_34_000_03: "I think it’s important for men to know that..."

A little bit about me

- 2.5 days here in clinical genetics
- 2.5 days in health promotion services (Solent NHS Trust)
- Final year of my professional doctorate in health psychology

What’s health psychology and what’s a professional doctorate?

- Health Psychology
  - Psychology of health and illness
- Professional doctorate
  - Equivalent in status and challenge to a PhD
  - Professional career
  - Qualify mid-2016
  - My skills

Key stages of Thematic Analysis

- Phase 1: Familiarising yourself with your data
- Phase 2: Generating initial codes
- Phase 3: Searching for themes
- Phase 4: Reviewing themes
- Phase 5: Defining and naming themes
- Phase 6: Producing the report

Masculinity/body image

- M_01_QUOTLE_10: "I probably see myself as being a role model, as being in a traditional sense and also kind of being in a environment where the idea of what a man is was quite narrowly defined as being kind of you know big and muscular and sort of..."
Feeling accepted

- M_40_GH_Luke_10: “I wanted to be a personal trainer, a P.E. teacher when I was attached, but they told me that my speech I wouldn’t be able to do that. And I could, I did train teachers when I was attached, I probably did teach under 8. And I always felt that I had a much better rapport with them than I did at school with them because we were all their age and they could relate and actually feel what it was like... I mean, I’m tall and it made it more real for them.”

Transition moments

- M_34_Todd_03: “I think, I missed out on my late 20s, making documentaries (posed) myself and new friends. It was very ambitious, very difficult projects, but after two years in the company of two other people, I think you know I learnt a huge amount about myself, and so there was a sort of subtext, that I think, eventually you get growing up, I think going back and writing a song and making a teacher at one of the toilets, and, and suddenly started a relationship, and it was like, "Christ, this easy!"”

Differences between participants

- M_19_Oll_05: “I’m going to start growing up around about that age and getting older. You start looking at yourself and asking whether you want to do, so, but for me it was a very different role than I didn’t have. I didn’t have to be a teacher when I was (posed) like it was a bit, and it’s amazing how I don’t know what world it is, it’s probably advanced in some way? Or disappointed in itself? Something like that. But not in a, just like sinh. I got this card.”

Confidence

- M_89_Oll_06: “It wasn’t comfortable, I didn’t think I could socialize with the people with this whole idea of girlfriends and all that stuff tending to be round about then and yes, probably a bit of fear or worry that I wasn’t going to cut it ever so.”

Time Scale

- September
  - Progression viva
  - January/February
  - Completed data generation (80 interviews)
  - June
- Initial analysis complete
Living with Russell-Silver syndrome: falling short?

What is Russell-Silver syndrome?

- Children are born with unexplained low birth weight. Growth is affected during childhood and results in below average height in adulthood (5’2” men/4’8” women).
- Body and face asymmetry.
- Children have a distinctive facial appearance.
- Treatment is growth hormone therapy (GH).
- In some people with RSS a genetic change is found: 40-50% are found to have a problem on chromosome 11 and in another 5-10% on chromosome 7, for the other 40% the cause is unknown.

Background

- Differing from the ‘norm’ could result in unpleasant feelings or emotions.
- Some people with a visible difference experience negative emotions, but many cope well with looking different.
- Severity is not related to level of psychological distress.
- Skills in resilience and coping are important.


Analysis

- Thematic analysis
  - Essentially a method for identifying and analysing patterns in qualitative data

Thematic Map

Results

'Symmetry's a good thing'

Oli (5’1”, 39 years old)
'Symmetry’s a good thing, it always has been. It’s the standard in everything else [...] that’s actually a big problem and people notice and that, in my later teen years really got to me, a lot.'
Results

Bits I don’t like

Glenn (5’, 69 years old)

- ‘I didn’t like the way I looked. I didn’t actually look at myself in the mirror with the ability to analyse very quickly what I exactly looked like, until I was probably about ten […] And I didn’t like what I was seeing. And so I could understand why the girls liked me for my personality, but short and ugly, I don’t think so.

Ideal man

Warner (4’9”, 37 years old)

‘Because girls are not going to find me attractive physically. I have to accept that […] because I’m, because of my height, I guess most girls are looking for guys who’re a bit taller. And my lack of muscle. Girls are looking for guys that have sort of got a bit more muscle.

Last word from Luke (4’8”, 40 years old)

‘If it should be more about being a big person in a little body you know what I mean, and then sort of you know who you are, what you stand for and how to cope with things’.

Results

‘Mayor of the friend zone’

Todd (5’6”, 34 years old)

‘I think one area where it did really did have an impact was with girls in that kind of teenage phase […] I think I had pretty low sort of sexual self-esteem, all the way through my teenage years and actually probably through most of my 20s […] I was very definitely kind of mayor of the friend zone […] I think I was always maybe slightly resentful that people would judge me very quickly on what I was’.

Conclusions

- Main findings
  - Adolescents, especially males, have appearance related concerns that impact on romantic relationships

- Application of this research
  - Psychological support
  - Health care professionals
  - Child Growth Foundation

With thanks to:

Dr Elizabeth Jenkinson (Dx)
Professor Angela Fenwick (Second supervisor)
Professor Karen Temple
Dr Kemi Lakulo-Soilpe
Dr Justin Davies
Dr Hazel Insik
Dr Deborah Mackay
Dr Chris Byrne
Dr Emma Winkel
Dr Renata Dias
Mrs Jenny Childs (Child Growth Foundation)
Living with Russell-Silver syndrome: falling short?

What is Russell-Silver syndrome?

- Children are born with unexplained low birth weight. Growth is affected during childhood and results in below average height in adulthood (5'2” men/4'8” women).
- Body and face asymmetry.
- Children have a distinctive facial appearance.
- Treatment is growth hormone therapy (GH).
- In some people with RSS a genetic change is found: 40-50% are found to have a problem on chromosome 7 and in another 5-10% on chromosome 7, for the other 40% the cause is unknown.

Background

- Differing from the ‘norm’ could result in unpleasant feelings or emotions.
- A preference for symmetrical faces may be related to sexual selection and mate choice in males and females and for symmetrical bodies and may be an indicator of health.
- Children with short stature (SS), compared to peers with normal stature, can often feel less satisfied with their appearance.
- Skills in resilience and coping are important.

Analysis

- Thematic analysis
  - Essentially a method for identifying and analysing patterns in qualitative data

Thematic Map

Results

“Symmetry’s a good thing”

Oli (5’1”, 39 years old)\
“Symmetry’s a good thing, it always has been. It’s the standard in everything else [..] that’s actually a big problem and people notice and that, in my later teen years really got to me, a lot.”
Results

Bits I don’t like
Glenn (5’, 69 years old)
• “I didn’t like the way I looked. I didn’t actually look at myself in the mirror with the ability to analyse very quickly what I exactly looked like, until I was probably about ten [...] And I didn’t like what I was seeing. And so I could understand why the girls liked me for my personality, but short and ugly, I don’t think so.”

Ideal man
Warner (4’9”, 37 years old)
 “[B]ecause girls are not going to find me attractive physically. I have to accept that [...] because I’m, because of my height, I guess most girls are looking for guys who are a bit taller. And my lack of muscle. Girls are looking for guys that have sort of got a bit more muscle.”

Last word from Luke (4’8”, 40 years old)
“[I]t should be more about being a big person in a little body you know what I mean, and then sort of you know who you are, what you stand for and how to cope with things.”

Results

‘Mayor of the friend zone’
Todd (5’6”, 34 years old)
“I think one area where it did really did have an impact was with girls in that kind of teenage phase [...] I think I had pretty low sort of sexual self esteem, all the way through my teenage years and actually probably through most of my 20s [...] I was very definitely kind of mayor of the friend zone [...] I think I was always maybe slightly resentful that people would judge me very quickly on what I was”

Conclusions

• Main findings
  - Adolescents, especially males, have appearance related concerns that impact on romantic relationships

• Application of this research
  - Psychological support
  - Health care professionals
  - Child Growth Foundation

With thanks to:
Dr Elizabeth Jenkinson (DoS)
Professor Angela Fenwick (Second supervisor)
Professor Karen Temple
Dr Kemi Lokulo-Sodipe
Dr Justin Davies
Dr Hazel Inskip
Dr Deborah Mackay
Dr Chris Byrne
Dr Emma Wakeling
Dr Renuka Dias
Mrs Jenny Childs (Child Growth Foundation)
Understanding the lived experience of Russell-Silver syndrome – recommendations for healthcare professionals

Lisa Marie Ballard\(^1\), Elizabeth Jenkinson\(^2\), Karen Temple\(^3\) and Angela Fenwick\(^3\)


**Background** - Russell-Silver syndrome (RSS) is a rare genetic condition characterised by slow pre and post-natal growth, short stature in adulthood, triangular facial appearance and body/facial asymmetry. Little is known about what it is like to live with this syndrome beyond what is reported in the clinical literature. Research shows that having a visible difference may result in psychological distress (Harcourt & Rumsey, 2012). A need for clear guidance for families and healthcare professionals (HCPs) in how to communicate and support these patients has been identified. Our research aim was to understand what it means to live with RSS.

**Methods** - Fifteen in-depth, semi structured interviews were conducted with participants from the UK aged between 25 - 69 (6 women) and thematic analysis was used to identify themes.

**Results** - Four themes were identified focusing on visible differences (height, face shape, asymmetry, low muscle mass) which impacted participant's confidence and self-esteem. Some participants remained resilient, others experienced psychological distress. These issues significantly influenced male participants ability to form romantic/intimate relationships in adolescence and adulthood.

- **It's not all about height** – issues other than height caused appearance related concerns.
  - ‘Symmetry’s a good thing, it always has been. It’s the standard in everything else [...] that’s actually a big problem and people notice and that in my later teen years really got to me, a lot’.

- **Being a big person in a little body** – confidence & self-esteem.
  - ‘It’s made me a more outgoing, confident person [...] you either speak up for yourself or you drown, it’s made me a more positive person, you let your views be known and you’re not letting people talk for you’.

- **Sharing, comparing & compensating** – dealing with visible difference in adaptive & mal-adaptive ways.
  - ‘I think it was nice knowing that you weren’t on your own and that there was other people like you, and that was good knowing that other people had symptoms similar to me’.

- **Mayor of the friend zone** – only establishing a friendships when a romantic relationship is desired.
  - ‘In that teenage phase [...] I was very definitely kind of mayor of the friend zone that I'd become very good friends with all of these very beautiful girls [...] I sort of really struggled with forming relationships’.

**Discussion** - The focus for HCPs involved in the care of children with growth problems is often on height. There is a clear treatment pathway using growth hormone therapy, but no robust evidence to support its effectiveness in children with RSS. Our research suggests patients struggle with varied psychosocial, appearance and body image related concerns which often overshadow a concern about height. Our recommendations for HCPs working with this patient group are:

- **HCPs should receive training to identify appearance related concerns.**
- **Create a specific measure to identify appearance related concerns in patients with growth problems.**
- **Evaluate the potential of psychosocial support/interventions.**

Lisa Ballard was funded by the UK National Institute for Health Research, Research for Patient Benefit programme (PB-PG-1111-26003), was supported by the NHRI Wessex CRN and this research partially fulfilled the requirements of the University of the West of England for the degree of Professional Doctorate in Health Psychology.
Falling short? The psychosocial impact of living with Russell-Silver syndrome

By Lisa Marie Ballard, Angela Fenwick, Elizabeth Jenkinson, and Isabel Karen Temple.

Russell-Silver syndrome (RSS) is a rare genetic condition that restricts growth and in many cases, causes asymmetry. There is little research exploring how this appearance-altering, genetic condition affects people’s psychosocial functioning. Lisa Marie Ballard et al review the existing literature on short stature and RSS to provide insights for health professionals, and inform ongoing and future research.

In a society that increasingly values appearance, anyone that differs from the ‘norm’ or what is culturally defined as ‘attractive’ may experience psychological distress based on the reactions and perceptions of others, as well as their own (Harcourt and Rumsey, 2012). However, people living with appearance-altering congenital conditions may not be receiving the psychological support they need to increase their confidence and self-esteem, and build resilience to develop and retain healthy romantic relationships in adolescence and into adulthood.

In this article, the authors discuss the potential impact that looking ‘different’ could have on people with Russell-Silver syndrome (RSS)—a rare genetic condition that affects height and facial/body symmetry. The authors will also outline a study being conducted by researchers in Southampton with adolescents and adults who have the condition.

‘Having a negative body image may result in lower self-esteem and in turn affect psychological wellbeing, causing anxiety and depression’

Russell-Silver syndrome

RSS is genetic, but not usually inherited (Binder et al, 2011). It is characterised by slow growth in the womb, poor postnatal growth, short stature in adulthood, triangular facial appearance with a broad prominent forehead, and body/facial asymmetry (Wakeling, 2011). The prevalence of RSS is not easily determined, as making a clinical diagnosis is difficult due to ill-defined features that vary in severity (Wakeling, 2011); the published incidence of RSS ranges from 1:3000 to 1:100 000 (Price et al, 1999). Males and females are equally affected (Binder et al, 2011).

At present, there are two known molecular abnormalities that cause RSS. First, a loss of DNA methylation at the H19 locus causes 60% of cases (Netchine et al, 2007). This epigenetic mutation causes a reduction in the growth-promoting factor, insulin-like growth factor 2. Second, maternal uniparental disomy of chromosome 7 (matUPD7)—the inheritance of both chromosome 7s from the mother with no contribution from the father—causes 5–10% of cases (Netchine et al, 2007). The remaining patients have clinical features of RSS, but an unknown molecular cause (Wakeling, 2011).

The primary treatment for RSS is growth hormone (GH) for short stature. While there is evidence that this increases height in some growth conditions (Dahlgren, 2011), it is less clear how effective it is for RSS. GH cannot treat other appearance differences, such as asymmetry, and might make them more prominent. Other options include procedures for
overcrowded teeth or leg-lengthening surgery.

Changes to appearance

RSS affects appearance in several ways, notably in short stature and asymmetry. Children often show slow growth throughout childhood, coupled with reduced growth at birth and typically no catch-up growth during puberty. This results in a height -4.2 standard deviations below the mean in adulthood, with an average height of 140 cm (4'7'') in women and 151 cm (4'11'') for men (Binder et al, 2011). To put this into perspective, the average height for men in the general population is 5'8'' and for women it is 5'4'' (Royal College of Paediatrics and Child Health, 2015). Asymmetry can affect the body, face and/or limbs and prevalence varies from about one-third upwards (Price et al, 1999; Wakeling et al, 2010).

Psychosocial impact of being visibly different

Being shorter than average may impact significantly on the development of self-esteem and body image in children and young people. Research has found that young adults with short stature, compared with peers within the normal range, rate themselves as less attractive (Cooke, 2004).

Short stature can predict negative body image and this is often found to be the case in boys more so than girls (Vilhjalmsson et al, 2012). Having a negative body image may result in lower self-esteem and in turn a decrease in psychological wellbeing, causing anxiety and depression (Vilhjalmsson et al, 2012). Problems can begin when an individual’s body image deviates from the cultural ideal; for example, being a taller man rather than shorter is seen as more desirable, as taller men may receive more favourable attention and therefore have more opportunities romantically and socially (Vilhjalmsson et al, 2012).

However, being short does not necessarily lead to negative psychosocial outcomes. Recent research published by Schanke and Thorsen (2015) identified coping strategies that can be used to deal with psychosocial challenges. Ten people aged 45–65 years old were interviewed and findings highlighted the important role of stigma-handling and resilience throughout the lifespan. The challenges and coping strategies of people with short stature are beginning to be understood; however, the complexities require further exploration to help support their needs.

Symmetry and beauty

A further societal pressure stems from the widely held perception that symmetry is linked to beauty. Patients with RSS often experience visible asymmetry; for example, one side of their body could be visibly smaller than the other, and weight gain may accentuate this difference further.

Evidence shows that a preference for symmetrical faces may be related to sexual selection and partner choice for both men and women (Thornhill and Gangestad, 1999; Rhodes, 2006; Little et al, 2008; Grey and Little, 2014). Having a symmetrical face may be viewed as an indicator of having healthy genes to pass onto offspring (Thornhill and Gangestad, 1999). Findings are similar for sexually dimorphic faces, i.e. women with feminine features and men with masculine features (Grammer and Thornhill, 1994;
Rhodes, 2006; Little et al, 2008; Grey and Little, 2014), and for symmetrical bodies (Gangestad and Simpson, 2000).

Having romantic relationships is one of the most important developmental experiences in adolescence, as they can improve self-esteem, enhance emotional support and enable individuals to form healthy relationships (Sorensen, 2007). However, evidence exploring the impact of asymmetry on psychosocial adjustment and intimacy for patients with RSS is lacking, and this area is also under-researched in patients with other appearance-altering conditions (Sharratt, 2015).

Enhancing treatment and psychosocial support

The Wessex Imprinting Group, based at the University of Southampton, is conducting a study with people who have RSS (STAARS UK—Study of Adults and Adolescents with Russell-Silver Syndrome). This study was initiated as The Child Growth Foundation, a UK charity for children and adults with growth and endocrine issues, has pressed for more information on health outcomes for adults with RSS, as little research exists.

The STAARS study has three main aims. First, the study seeks to ascertain what the long-term health outcomes are for people with RSS, as there is significant evidence to link the condition with an increased risk of heart disease (Barker et al, 1989; Hales et al, 1991; Barker et al, 1993; Osmond et al, 1993; Barker, 2005). It has also been found to be associated with diabetes in adulthood (Hales et al, 1991; Barker et al, 1993; Barker, 2005). The study’s second aim concerns GH, the main treatment offered to children and adolescents with RSS. Finally, the study aspires to involve exploring the lived experience of individuals through in-depth interviews with a subset of the total sample.

Researchers are looking to recruit 100 people with RSS syndrome to undertake clinical investigations to understand more about health outcomes.

Conclusion

People with RSS are living with a visibly different appearance and research shows looking different from the ‘norm’ may result in psychological distress. The in-depth interview aspect of the STAARS study may give an insight into how people with RSS feel about their appearance and what impact looking different has on them and their relationships.

Children and adolescents are not routinely screened for psychological issues or offered psychological support for appearance-related concerns in endocrine clinics, as clinicians generally focus on treating growth. All health professionals working with patients who have an appearance-altering condition should be aware of the possible psychosocial impact on the development of social, and especially romantic, relationships.

Key points

- Having an appearance that differs from the ‘norm’ can negatively impact on a person’s wellbeing and some people are more resilient than others
- Forming romantic relationships is an important part of development during adolescence and those with Russell-Silver syndrome (RSS) may find this harder
Health professionals working with adolescents with appearance-altering conditions, such as RSS, and other growth disorders, are in a position to ask questions about how their appearance affects different aspects of their lives.

The STAARS study will report its findings in 2016/2017. To find out more about the outcomes, please visit: www.southampton.ac.uk/geneticimprinting/informationpatients/staars.page

Acknowledgements: Lisa Ballard was funded by the UK National Institute for Health Research (NIHR) Research for Patient Benefit programme (PB-PG-1111-26003), and was supported by the NIHR Clinical Research Network Wessex.

References


Overview of the study
Medical study (Dr. Farn Sadagh)
Aims
- What are the long-term health outcomes?
- How does growth hormone influence height?
Recruitment
- 28 adults and adolescents

Overview of the study
Qualitative study
Aims
- What is it like to live with RSS?
Recruitment
- 20 adults and adolescents

Early findings
You don’t stop having RSS once you reach your final height
- Differences between men and women
- Frustration with non-specialist health professionals
- Uncertainty over what symptoms are related to RSS

Early findings
It’s not all about height
- Height is not the only issue
- Some people have issues with the way they looked
- Differences between men and women

Feedback
Who needs to know about these findings?
Is there anything that could have made a difference for you during adolescence?
<table>
<thead>
<tr>
<th>Date</th>
<th>Type</th>
<th>Name</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>03/12/2014</td>
<td>Supervision</td>
<td>AF</td>
<td>Starting new job</td>
</tr>
<tr>
<td>08/12/2014</td>
<td>Supervision</td>
<td>EJ</td>
<td>Discussed new supervisory team whilst Liz is away</td>
</tr>
<tr>
<td>08/12/2014</td>
<td>Meeting</td>
<td>AF, KS, KT</td>
<td>Introductions to new job</td>
</tr>
<tr>
<td>10/12/2014</td>
<td>Supervision</td>
<td>AF</td>
<td>Discussed RD1</td>
</tr>
<tr>
<td>11/12/2014</td>
<td>Supervision</td>
<td>EJ</td>
<td>Liz Suggested using TA rather than IPA, recruiting only adults for my thesis sample</td>
</tr>
<tr>
<td>06/01/2015</td>
<td>Supervision</td>
<td>EJ</td>
<td>Liz was happy with my RD1 and happy to support me as a supervisor</td>
</tr>
<tr>
<td>12/01/2015</td>
<td>Supervision</td>
<td>AF</td>
<td>Been to RSS clinic with Karen, Kemi and Justin, attended participant visit with Kemi, adding to my literature review, started to look at Turner syndrome, Liz happy with RD1, James still not replied, how to keep track of recruitment, only attend Wednesday meeting when necessary, check with Kemi about NRES and musketeer, need a black CV to fill in for site file</td>
</tr>
<tr>
<td>16/01/2015</td>
<td>Supervision</td>
<td>JBD</td>
<td>James - happy with RD1, ambiguity in intro</td>
</tr>
<tr>
<td>19/01/2015</td>
<td>Present to team</td>
<td>KS, KT, AF</td>
<td>Present my progress so far</td>
</tr>
<tr>
<td>30/01/2015</td>
<td>Steering group meeting</td>
<td></td>
<td>Discussed presentation for UWE about research progress</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Need to develop a buddy system</td>
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<td></td>
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<td></td>
<td>Can I get a safe stick?</td>
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<tr>
<td></td>
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<td></td>
<td>Piloting – who will it be, don’t pitch it like a pilot</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Send over what I have done so far around the life grid</td>
</tr>
<tr>
<td>05/02/2015</td>
<td>Supervision</td>
<td>AF, EJ</td>
<td>James February deadline is not realistic, 15 participants for professional doctorate thesis is more than enough.</td>
</tr>
<tr>
<td>10/02/2015</td>
<td>Supervision</td>
<td>AF</td>
<td>Instructions for transcriber</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td>Reflection for both interviews</td>
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<td></td>
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<td></td>
<td>Transcription, it’s not conversational analysis</td>
</tr>
<tr>
<td>03/03/2015</td>
<td>Supervision</td>
<td>AF</td>
<td>I asked for feedback on my interviewing/questions.</td>
</tr>
<tr>
<td>10/03/2015</td>
<td>Supervision</td>
<td>AF, JBD</td>
<td>I updated them both on my progress, discussed progression report deadline of June, and discussed internal examiners for progression viva.</td>
</tr>
<tr>
<td>02/04/2015</td>
<td>Supervision</td>
<td>AF</td>
<td>Discussed RD1 outcome, write piece for CELS page, research questions, don’t mention clinical arm too much</td>
</tr>
<tr>
<td>19/05/2015</td>
<td>Supervision</td>
<td>AF</td>
<td>Feedback on interviews and transcripts (Q – Darin’s occupation seems quite identifiable), ethics of change in recruitment protocol, change of desk, meant to have skype with James and AF – didn’t happen, look up narrative analysis, feedback contextual information about RSS, Yin – Case Studies</td>
</tr>
<tr>
<td>10/06/2015</td>
<td>Meeting</td>
<td>KT, KS</td>
<td>Discussed making an amendment if asking if RSS people want to just take part in qualitative, progress with recruitment, thank you letters, still recruit to qualitative study but ask if we can take basic measurements</td>
</tr>
<tr>
<td>24/06/2015</td>
<td>Supervision</td>
<td>AF</td>
<td>My supervisor doesn’t like the term life span ‘how does the lived experience of having RSS change across the life span’ I have spent some time thinking of alternatives and I think it just comes down to sociologists who use life course and psychology using lifespan. I am going to keep lifespan.</td>
</tr>
<tr>
<td>26/06/2015</td>
<td>Steering group</td>
<td></td>
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</tbody>
</table>
24/07/2015  Supervision  AF  Confirming arrangements for progression exam.

18/08/2015  Supervision  AF, JBD  We discussed me taking out the use of case studies in my project as it wasn’t necessary and is more than I need to do to complete what is required of me. James gave me feedback on my latest draft of my progression report (tell a story for my intro, clinical features in a table, included info sheet and consent forms in appendix), I must submit my UWE ethics application.

14/09/2015  Meeting  KT, KS  Conference – present to adults, get a room to do interviews and medical exams, present at 7th Oct teaching session.

25/09/2015  Steering group meeting  I presented some for my developing themes - they were surprised by the effect asymmetry has on participants, they want me to include height in my participant identifier, lit review = what is known, what is not known, why is that important.

06/10/2015  Supervision  AF  Analyse three transcripts together. Get a timetable together for Angela and Liz and include when I hope to finish
Set up a skype meeting with Angela and Liz (26/10/2015?) Ask Liz does my word count include quotes? Revisit interview schedule in light of developing themes. Pick one developing theme and tell its story. Focus on what stories I am trying to tell (parents, psychologist, clinicians, not fitting in with medical model). Think about analysing and verifying at the same time. Start to plan thesis.

12/10/2015  Meeting  KT, KS  Went through all the participants Kemi has seen and all the ones I have seen and contacted. AF asked me what the disparity was between who Kemi has seen (28) and how many I had seen and anticipate seeing. This activity really helped as I realise that four of Kemi’s don’t have a genetic diagnosis of RSS and four to five are under 18. I will contact them when I have recruited the 15 I need for my thesis.

16/10/2015  Supervision  AF, EJ  I updated Liz on my progress to date, asked about my workplace supervisor as mine has left, Liz will send me some electronic theses as examples so I can work out a realistic timescale for submission, we discussed meeting face to face on the 30th November.

26/10/2015  Supervision  AF, EJ  I updated Liz on my progress to date, asked about my workplace supervisor as mine has left, Liz will send me some electronic theses as examples so I can work out a realistic timescale for submission, we discussed meeting face to face on the 30th November.

17/11/2015  Meeting  KT, KS  Whilst going through one of the transcripts and the participant was talking about weight I wondered is this to do with RSS? So I have a meeting with Kemi and Karen and they are really interested in why some of the RSS patients put on weight and some stay slim. It seems as if it is related in some way, the classic RSS patient does put on weight in adulthood and is part of the RSS experience.

30/11/2015  Supervision  AF, EJ  Developing theory - making sense of what I’ve found and what participants have said to me
Time scale - don’t start with lit review as it will need more work once the analysis is done, I could collate tables of lit and what each study found, ready to go into the lit review. Lit review is the background to my findings. I need my thesis to be tight and precise.
<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Notes</th>
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<tbody>
<tr>
<td>18/12/2015</td>
<td>Steering group meeting</td>
<td>When mentioning how asymmetry was more common a problem, the clinicians (Justin - paediatric endocrinologist) said how they were unaware this plays such a big problem and they would just concentrate on height without really considering there are other issues. I asked if psychological support is offered routinely and it is not. It really feels as if I am uncovering issues that are of real value to the clinicians and opening their eyes to what their patients are going through. Justin said that he just notes the characteristics and then just carries on without thinking about how they will affect the individual later on in life. Body fat and asymmetry - I asked the question about how some of the participants have said that gaining weight has highlighted their asymmetry more and is there a reason for this. They said they did not know and that that could be an interesting aspect to look at. We could compare the scans with what the participants say about themselves.</td>
</tr>
<tr>
<td>18/01/2016</td>
<td>Supervision</td>
<td>I had a meeting with AF to discuss my coding so far. She was happy with what I have done and suggested that often it's more about chunking data together and then looking at what you have in those chunks to refine the data. My codes are a bit more detailed than that but not too detailed. In speaking with AF we started to see some links between some of the codes which led me to make this diagram. The codes under coping I had grouped together, but they didn't have a theme, and then once I thought about them I decided they were mostly about coping.</td>
</tr>
<tr>
<td>09/02/2016</td>
<td>Supervision</td>
<td>Should I apply to present research at the European Health Psychology conference in Aberdeen? Overview of recruitment.</td>
</tr>
<tr>
<td>16/02/2016</td>
<td>Supervision</td>
<td>Going to go through the bigger codes to see if they themes themselves or need to be broken down into smaller codes. Do I have some overarching themes? General (+ lifespan), male and female?</td>
</tr>
<tr>
<td>14/03/2016</td>
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<td>Date</td>
<td>Event</td>
<td>Participants</td>
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<tr>
<td>05/04/2016</td>
<td>Supervision</td>
<td>AF, KT</td>
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<td>26/04/2016</td>
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<td>Participants</td>
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<td>27/09/2016</td>
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<td>06/10/2016</td>
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<tr>
<td>24/10/2016</td>
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<tr>
<td>15/11/2016</td>
<td>Supervision</td>
<td>AF, EJ</td>
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Appendix 16 – Systematic review ‘Increasing the uptake of stop smoking support in secondary care: A systematic review’

Deadline 04 Aug 2014 14:00

Coursework Item SYSTEMATIC REVIEW (USPJHKH-30-M) CW1 WRITTEN REPORT OF THE SYSTEMATIC REVIEW (6000 WORDS) (CW1)
Table of contents

Abstract .................................................. 3
Introduction ............................................. 3
Objective .................................................. 4
Methods ................................................... 5
  Protocol .................................................. 5
  Report eligibility criteria ............................ 5
  Study eligibility criteria ............................. 5
    Types of studies .................................... 5
    Types of participants ............................... 5
    Types of interventions .............................. 5
    Types of comparisons ............................... 6
    Types of outcome measures ....................... 6
  Search methods for identification of studies ....... 6
    Electronic searches .................................. 6
    Searching other resources ......................... 6
Data collection and analysis .......................... 6
Selection of studies .................................... 7
Quality assessment ...................................... 7
Data extraction and management ....................... 8
Risk of bias in individual studies .................... 8
Summary measures ....................................... 9
Synthesis of results .................................... 9
Risk of bias across studies ........................... 9
Additional analysis ..................................... 10

Results .................................................. 11
Study selection .......................................... 11
Study characteristics ................................... 12
Methods .................................................. 12
Participants ............................................. 13
Intervention ............................................. 13
Outcomes ................................................. 1
  Risk of bias within studies ......................... 17
  Results of individual studies ....................... 18
  Synthesis of results .................................. 20
  Narrative synthesis ................................... 2
    Preliminary synthesis ................................ 20
    Exploring relationships ............................. 2
      Within studies .................................... 21
      Between studies ................................... 21
    Narrative synthesis by subgroup .................. 2
Discussion ............................................... 2
Summary of evidence .................................... 26
Limitations .............................................. 2
  Outcome level limitations ............................ 27
  Study level limitations ............................... 28
  Review level limitations ............................. 28
Conclusion .............................................. 28
Funding .................................................. 29
Word count ............................................... 29
References ............................................... 29
Appendix 1 ............................................... 32
<table>
<thead>
<tr>
<th>Appendix 2</th>
<th>33</th>
</tr>
</thead>
<tbody>
<tr>
<td>Appendix 3</td>
<td>34</td>
</tr>
</tbody>
</table>
Increasing the uptake of stop smoking support in secondary care: A systematic review.

Lisa M Hodges & Nisha Sharma (Solent NHS Trust)

Abstract

Objectives: The main objective was to determine the most effective method of increasing the uptake of stop smoking support by smokers in the secondary care. Data sources: EMBASE MEDLINE, PsychINFO, CINAHL, Social Science Citation Index, and the Cochrane Library and Campbell Collaboration databases were all searched. Hand searches on reference lists were also conducted. Study eligibility criteria: Only randomised controlled trials were used in this review. Participants: Patients and visitors to secondary care who were over 18 years of age. Synthesis methods: Narrative synthesis. Results: Seven studies were included in this review. Interventions from 30 seconds to 60 minutes made significant differences in abstinence or smoking cessation support attendance. Also, fax referrals to cessation services and follow-up telephone calls were effective in increasing the uptake of smoking cessation support in secondary care. Limitations: Studies were very heterogeneous in their robustness. Limitations of this review were a limiter in the search strategy to find only English language articles and unpublished data was not successfully obtained. Conclusion: These results are of particular importance to clinical care leads, commissioners and providers to inform smoking interventions in secondary care. Recommendations for future research are made.

Introduction

Smoking kills approximately half of all smokers which results in around 100,000 people in the UK every year and continues to be the leading cause of premature death. One third of all respiratory deaths are smoking related as are a quarter of all cancer deaths and about 1 seventh of all cardiovascular deaths (ASH, 2014). NICE guidance (2008) recommended the use of brief advice, one-to-one counseling, group support and pharmacotherapy as proven interventions to help smokers quit and Rigotti, Clair, Munafo & Stead (2012) in a review concluded that intensive behavioural support programmes that begin in hospital and carry on after discharge promote smoking cessation regardless of diagnosis. Providing secondary care patients information and support to stop smoking will have an impact on survival rates, complications, fewer bed days, wound healing, drug doses and hospital readmission rates (NICE guidance 48, 2013). It is therefore health care professionals duty to their patients in secondary care to give access to smoking cessation support. The NICE guidelines from November 2013 highlighted gaps in the evidence around effectiveness of interventions to increase the uptake of smoking cessation support and also made recommendations for research in this area. It was the NICE guidelines that informed this systematic review question.

This is a new review with the main aim being to build on the knowledge that smoking interventions that begin in secondary care promote cessation and expand the topic by asking which are the most effective interventions that increase engagement and uptake of smoking cessation support in secondary care. The outcomes of this review are of importance and interest to those who commission smoking cessation services as well as those who provide this support, clinical care leads in secondary care and any healthcare staff working in secondary care. The review outcomes are also of importance to the smokers and their families who may benefit from support whilst visiting secondary care services.

The current knowledge in this area is from NICE guidance (2013) which states that in the UK all secondary care providers should be identifying smokers as soon as possible upon admission and offering them pharmacotherapy and intensive behavioural support to be smokefree during their stay at least and to follow patients up at discharge to monitor longer term abstinence. However, the practical challenges of this recommendation have not been wholly overcome (Murray, 2013).
This review aims to address the gap in the evidence between smokers in secondary care and the highly effective stop smoking interventions. It hopes to answer how, when and where should health care professionals working in secondary care identify and engage their smoking patients with the smoking cessation support service?

Objective
The main objective was to determine the most effective method of increasing the uptake of stop smoking support by smokers in the secondary care setting by reviewing randomised, controlled trials.

Methods
Protocol
The protocol for this study can be found in appendix 1.

Criteria for considering studies for this review
Report eligibility criteria
No eligibility criteria was set around length of follow-up or year of publication, but search criteria was set to look for human, adult studies. We also set the search to look for English language articles only as there were no funds or facilities to translate articles. There was no need to include any 'abstract only' studies as all abstracts selected for inclusion had full text articles available.

Study eligibility criteria
Types of studies
We included randomised controlled trials only (one study, Murray 2013, was a cluster randomised controlled trial).

Types of participants
We included studies who's participants were patients or visitors to a secondary care setting (including outpatient clinics, emergency departments, could include staff, but not exclusively staff) and were over 18 years of age. We excluded medical settings like primary care and walk-in centres and excluded patients with mental health issues, drug or alcohol dependency and those who were pregnant.

Types of interventions
We included studies which focused on the identification of smokers, recruitment of smokers into a smoking cessation program, engagement around smoking cessation and patients/visitors, very brief advice, staff attitudes, patient/visitor attitudes, and the provision of nicotine replacement therapy (NRT) and behavioural support. We excluded studies which solely reported on attitudes and therefore were not interventions and we also excluded studies that were comparing interventions, for example, different forms of pharmacotherapy, as these have been reported in other reviews (Rigotti et al. 2012).

Types of comparison
We included studies that compared an intervention to either a control condition or routine/usual care.

Types of outcome measures
We included studies that reported on referral rates, engagement in the community smoking cessation service and provision of NRT. As a secondary outcome we included studies that also reported on quit rates, but only if that study reported on one of the primary outcomes also. We had no exclusion criteria for how abstinence or 7 day point prevalence was reported as self-report is the generally reported measure.

Search methods for identification of studies
Electronic searches
We devised search strategies and LH ran electronic searches in EMBASE (appendix 2), MEDLINE, PsychINFO, CINAHL, Social Science Citation Index, and the Cochrane Library and Campbell Collaboration databases. The most recent search was run on the 29th March 2014.

Searching other resources
We emailed the Cochrane Tobacco Addiction Group to check if any similar reviews were being conducted and there were not. LH hand searched for trials by checking reference lists, but did not find any additional trials to add to the review. We contacted authors for any missing data or for any part of the data extraction process that was deemed ‘unclear’.

Data collection and analysis
The following search terms were used for the database searches (full search strategy in appendix 2): smoking cessation programme; nicotine; tobacco; quit*; smok*; stop; cess*; giv*; engage*; motivat*; incentive*; intention; interact*; promot*; recruit*; self-selection; volunteer; invit*; enrol*; entry; patient participation; uptake; patient; smoker; consumer; service user; customer; client; hospitalization; outpatient*; secondary care; speciali#ed care; acute; hospital*; intervention; randomi#ed controlled trial*.

Selection of studies
The search strategies were implemented by LH and 7,253 studies were found (excluding duplicates). These titles were screened for eligibility and 105 titles were identified as potential trials to be included in the review (see figure 2 for flow diagram). The main reasons for removal of title, other than them being completely irrelevant, were primary care settings, focus on cost effectiveness, specific groups i.e spinal cord veterans, mental health or maternity settings and studies about harm reduction. Both LH and NS used the exclusion and inclusion criteria to grade each abstract. Once the abstract grading was compared there were some discrepancies which were mainly due to either oversight or differences in interpreting the trials. These were resolved and 9 articles were found to meet all the protocol requirements.

Quality assessment
Once the full texts were obtained for all 9 trials both LH and NS used the quality assessment tool for quantitative studies developed by the Effective Public Health Practice Project (EPHPP), McMaster University. It was during this process that two of the trials were excluded (see flow diagram, figure 2, for reasons). The results of both reviewers were compared and there were some discrepancies which were due to oversights (see table 1 for quality assessment outcomes).

Table 1. Outcomes by study of the Quality Assessment Tool for Quantitative Studies

<table>
<thead>
<tr>
<th>Report ID</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Selection Bias</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
</tr>
<tr>
<td>Study Design</td>
<td>Strong</td>
<td>Strong</td>
<td>Strong</td>
<td>Strong</td>
<td>Strong</td>
<td>Strong</td>
<td>Strong</td>
</tr>
<tr>
<td>Confounders</td>
<td>Strong</td>
<td>Strong</td>
<td>Strong</td>
<td>Weak</td>
<td>Strong</td>
<td>Strong</td>
<td>Strong</td>
</tr>
<tr>
<td>Blinding</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Strong</td>
</tr>
<tr>
<td>Data Collection Methods</td>
<td>Strong</td>
<td>Weak</td>
<td>Moderate</td>
<td>Strong</td>
<td>Strong</td>
<td>Weak</td>
<td>Moderate</td>
</tr>
<tr>
<td>Withdrawals and drop-outs</td>
<td>Weak</td>
<td>Strong</td>
<td>Moderate</td>
<td>Strong</td>
<td>Weak</td>
<td>Moderate</td>
<td>Weak</td>
</tr>
<tr>
<td>Was consistency of intervention measured?</td>
<td>Can’t tell</td>
<td>Can’t tell</td>
<td>Can’t tell</td>
<td>Can’t tell</td>
<td>Yes</td>
<td>Can’t tell</td>
<td>Can’t tell</td>
</tr>
</tbody>
</table>
Data extraction and management
One reviewer extracted the data using a form based on the Cochrane Intervention Review form - RCT’s V2, January 2013 (See appendix for an example). As reported above, missing data was requested from study authors.

For each study the following data were extracted:
- Study aim, design and unit of allocation
- Duration of participation, population, setting and method of recruitment
- Withdrawals, exclusions and missing data
- Baseline imbalances
- Description of intervention and control conditions and numbers allocated to each
- Primary and secondary outcomes were described and reported
- Author conclusions and funding sources.

Risk of bias in individual studies
One reviewer also reported on the risk of bias using the Cochrane risk of bias tool in the data extraction form, the results of this assessment are in figure 1 and table 2. No trials were removed from the analysis at study level due to risk of bias.

Figure 1. Risk of bias graph: review authors’ judgements about each risk of bias item presented as percentages across all included studies.

Summary measures
The principle summary measures for dichotomous outcomes such as self reported abstinence (Murray 2013 also used expired CO) at a variety of follow-up times (discharge, 4, 6, 12 weeks, 6 and 12 months) were odds ratios (but only for three studies, the other four used percentages), percentage of in-patients offered and accepted pharmacotherapy and behavioural support, percentage of patients discharged with pharmacotherapy and behavioural support arranged, number of quit attempts post intervention, sustained abstinence, attendance of community support post intervention, and contacted stop smoking service post intervention. The principle
Summary measures for continuous outcomes such as intervention time, number of cigarettes smoked daily and level of readiness to quit were differences in means.

Synthesis of results
One of the studies (Lewis 2009) used three groups, one control and two intervention conditions. In the results we compared the control and the third intervention group to make all seven studies homogenous. Also, some studies used slightly different follow-up points, these were adjusted and combined to increase homogeneity.

Risk of bias across studies
Risk of bias was found in three of the seven studies included in this review (see figure 1 and table 2). Richman 2000 did not use intention to treat analysis in their study to adjust for lost to follow-up participant outcomes. This has the potential to increase positive bias in outcomes. Murray 2013 could not randomly allocate their participants to intervention or control conditions as a cluster design to allocate 18 wards to condition was used, although this was unlikely to have had a significant effect as patients are allocated by other methods to wards. And lastly Lin 2013 had a bias concerning selective outcome reporting as they reported an outcome in the results that was not mentioned in the method section. As we did not have access to the protocol, there is a possibility this outcome was missed from the methods section, but was reported in the protocol. This outcome would have particular bearing on one outcome and that is of abstinence or point prevalence data. All seven studies were very robust in their blinding of outcome assessors and random sequence generation.

Table 2. Risk of bias assessment outcomes

<table>
<thead>
<tr>
<th>Report ID</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Random sequence generation</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
</tr>
<tr>
<td>Allocation concealment</td>
<td>Low</td>
<td>Low</td>
<td>Unclear</td>
<td>High</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
</tr>
<tr>
<td>Blinding of participants and personnel</td>
<td>Low</td>
<td>Low</td>
<td>Unclear</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
</tr>
<tr>
<td>Blinding of outcome assessment</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
</tr>
<tr>
<td>Incomplete outcome data</td>
<td>Low</td>
<td>Low</td>
<td>High</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
</tr>
<tr>
<td>Selective Outcome reporting</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>High</td>
<td>Low</td>
</tr>
<tr>
<td>Other bias</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
<td>Low</td>
</tr>
</tbody>
</table>

Additional analyses
No additional analyses were performed for this review.

Results
Study selection
Seven studies reporting, identifying or recruiting smokers in secondary care were included in this synthesis (see figure 2 for flow diagram). We searched MEDLINE, PsychINFO, CINAHL, EMBASE, Social Science Citation Index, Cochrane Library and Campbell Collaboration as well as hand searching relevant studies reference lists and contacting researchers and found a total of 9,606 records (9,595 were from electronic databases). After screening titles we found 105 potential abstracts that were reviewed by both reviewers to rate them against the study protocol, nine trials were found to meet all the criteria and full texts were found. Once these nine had gone through quality assessment it was found that two were not to be included as one reported on a smaller aspect of a main trial already included in the final papers and the second was a study protocol of a trial not yet completed. The main author of the study protocol was contacted to enquire if any data was ready to be released, but no further contact was received.

Study characteristics

Table 3 Summary of study characteristic

<table>
<thead>
<tr>
<th>Report ID</th>
<th>Author</th>
<th>Year</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Anders 2011</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>Lewis 2009</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>Richman 2000</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>Murray 2013</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>Warner 2011</td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>Lin 2013</td>
<td></td>
</tr>
<tr>
<td>7</td>
<td>Mahabee-Gittens 2008</td>
<td></td>
</tr>
</tbody>
</table>

Figure 2. Study flow diagram

<table>
<thead>
<tr>
<th>Design</th>
<th>RCT</th>
<th>RCT</th>
<th>RCT</th>
<th>RCT</th>
<th>RCT</th>
<th>RCT</th>
</tr>
</thead>
<tbody>
<tr>
<td>Population</td>
<td>Patients in ED non-urgent care</td>
<td>Consecutive smokers aged 18 years or older, attending the hospital.</td>
<td>All medically stable, orientated patients 18 years and older who presented to the ED.</td>
<td>All smokers who reported that they were current smokers, or had smoked within four weeks of admission, to any of 18 medical wards</td>
<td>Patients in preparation for elective surgery.</td>
<td>Male outpatients who were current smokers attending clinics</td>
</tr>
<tr>
<td>Setting</td>
<td>ED</td>
<td>Two U.K hospitals</td>
<td>ED</td>
<td>A large teaching hospital in the UK</td>
<td>PreOperative Evaluation Center (POE)</td>
<td>Large modern general hospital</td>
</tr>
<tr>
<td>Total randomised no.</td>
<td>221</td>
<td>450</td>
<td>152</td>
<td>493</td>
<td>300</td>
<td>126</td>
</tr>
<tr>
<td>Duration of participation</td>
<td>3 months</td>
<td>13 months (55 weeks)</td>
<td>3 months</td>
<td>6 months</td>
<td>3 months</td>
<td>12 months</td>
</tr>
<tr>
<td>Duration of treatment (mean)</td>
<td>3.3 minutes</td>
<td>A = 20 mins B = 3 hrs C = 3 hrs</td>
<td>Not reported</td>
<td>Dependent on hospital stay</td>
<td>5 minutes</td>
<td>30 seconds</td>
</tr>
<tr>
<td>Frequency of treatment</td>
<td>One-off</td>
<td>A = one-off B &amp; C = intervention plus 3 weekly sessions</td>
<td>One-off</td>
<td>Dependent on hospital stay</td>
<td>One-off</td>
<td>One-off</td>
</tr>
</tbody>
</table>

**Methods**
Seven remaining trials, which were published between 2000 and 2013, had sample sizes ranging from 126 to 493. Four trials took place in the USA, two in the UK (one study conducted the trial in two hospitals) and one in China. Three trials recruited patients specifically from the emergency department (ED), two recruited patients from all over the hospital, one from outpatients and one recruited patients who were undergoing elective surgery. Five studies recruited adult smokers, over 18 years, male and female, one recruited just male patients and one trial recruited the smoking parents of children admitted to non urgent ED.

**Participants**
The seven studies included 2,101 participants who were all aged 18 or over and had been recruited from secondary care and were smokers.

**Intervention**
All interventions were comparing methods of engaging smokers in a stop smoking intervention and comparing this against either a control condition or usual care. Table 4 summarises each intervention.

**Table 4. Key elements of each intervention**

<table>
<thead>
<tr>
<th>Report ID</th>
<th>Author</th>
<th>Intervention</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Anders 2011</td>
<td>Fax referral to cessation service + follow-up calls - All non urgent patients who presented to the ED were referred to the advanced practice nurse who assessed the smoking status of all patients the advanced practice nurse provided strong and personalized advice to quit. Participants were then randomly assigned to either the intervention or control groups. Participants in the intervention group were asked if they were willing to have their contact information faxed to the cessation services, with the understanding that the faxed referral would generate a number of telephone calls from the treatment services.</td>
</tr>
</tbody>
</table>
Sixty min intervention + three more session + pharmacotherapy + appointment with community-based service + follow-up if did not attend - Patients received an intensive one-to-one session with the HSCS lasting 60 minutes. They were then given the same five leaflets given to Group B. The HSCS recommended that participants use NRT or bupropion, which was prescribed by their ward doctor or general practitioner. Three more sessions were arranged at weekly intervals with the HSCS, each lasting 25–30 min. After 4 weeks, participants were given a specific appointment to attend the community-based service within 7 days. If the smoker did not attend the community-based service within 7 days, the service sent one reminder letter followed by a single phone call 10 days later.

Standardised, scripted counseling from physician + written and oral referral to cessation service - Patients were identified by a previously validated question - "Do you smoke cigarettes now?". They received a two page pamphlet "Stop Smoking" (American Heart Association) and standardised scripted counseling including a written and oral referral to a smoking cessation program. The smoking cessation program consisted of one week of intensive motivational and educational sessions. Program attendees also had the opportunity to start nrt of bupropion.

Brief advice + intensive bedside support + prescription of NRT - All patients well enough to engage in discussion were given brief advice to quit, and offered help to do so, by the research team. The brief advice included an explanation of the benefits of quitting, the nature of the support available (tailored support, one to one counselling or behavioural support, and pharmacotherapy while in hospital and continued support after discharge), and an assessment of desire to receive support. Patients were provided with standard written information from the UK’s health service about smoking cessation, if interested. Those who accepted cessation support were visited at the bedside by one of three smoking cessation practitioners. The smoking cessation practitioners offered one to one counseling, to be delivered daily throughout admission (or as often as was acceptable to the patient). In the absence of contraindications, they prescribed dual nicotine replacement therapy.

Brief advice + description of cessation service + brochure + option of referral - The intervention included the following: (1) advice to quit smoking for as long as possible before and after surgery, with emphasis that "fasting" (i.e., abstinence) from cigarettes the morning of surgery was of particular importance; (2) a description of quitline services; and (3) distribution of a brochure that included the dedicated quitline telephone number that the subject could call to initiate a consultation and the option of a faxed referral to the quitline from the provider.

Thirty second warn about 1/2 deaths, advice to quit and refer - For the intervention group, the physician said the following in a standardized manner: 'From medical research, one out of two smokers will be killed by smoking. Smoking is harmful to your health, so you must quit immediately for your health. The smoking cessation clinic is open every Friday afternoon. Please attend the clinic as soon as possible.' This intervention 'WAR': Warn about 1/2 deaths, Advise to quit and Refer took <30 s (and could be done quickly in about 20 s). The control group did not have any intervention, which was the usual practice. No intervention was given at follow-up, which resembled the real world practice.

Ten-15 minute counseling session + referral to cessation service - Participants randomized to the Intervention group were given a brief (10- to 15-minute) counseling session by the principal investigator or trained clinical research coordinator while their child was waiting to be evaluated by the emergency department physician. Participants were encouraged to quit smoking and assessed for level of readiness to quit. Participants responding positively to the question: "Would you seriously consider quitting in the next 6 months?" were given a brief description of the Ohio Quitline, and then asked about their interest in being referred to the Quitline. This time period was chosen to encourage as many participants as possible to speak with Quitline counselors trained in increasing motivation to quit. For those participants not ready to quit in the next 6 months, study personnel used the 5Rs counseling technique to increase motivation (Fiore, et al., 2000). Then study personnel described the services offered by the Quitline and offered participants the opportunity for faxed Quitline referral. All participants not wishing to be contacted by the Quitline were offered written Ohio Quitline tobacco cessation brochures.

Outcomes
All seven studies had prioritised their outcome measures (primary/secondary) differently, but there were commonalities within the reported outcomes (see table 5). Four studies (Murray 2013, Warner 2011, Lin 2013 and Mahabee-Gittens 2008) reported a four-week point prevalence outcome of smoking cessation, although Mahabee-Gittens 2008 used a 6 week follow but we made the decision to include it with the other articles. Lin 2013 was the only study to measure abstinence at four weeks, which means participants reporting having not smoked at all from intervention to follow-up. Five studies (Anders 2011, Lewis 2009, Warner 2011, Lin 2013 and Mahabee-Gittens 2008) reported three month point prevalence outcomes for smoking cessation. Warner 2011 and Lin 2013 both also reported abstinence outcomes at three months. Murray 2013 and Lin 2013 reported on point prevalence and abstinence at six months. Lewis 2009 and Lin 2013 reported at 12 months for point prevalence, although Lewis 2009 was actually 55 weeks, it
was decided by the reviewer to include as 12 months. Lin 2013 reported abstinence from smoking at 12 months. Five studies (Anders 2011, Lewis 2009, Richman 2000, Murray 2013 and Lin 2013) reported outcomes for participants attending for smoking cessation support. Other outcomes reported included patient contacted service via telephone at three months (Richman 2000 and Warner 2011) and reduction of smoking at three months (Lin 2013 and Mahabee-Gittens 2008).

Table 5 All outcomes for all studies

<table>
<thead>
<tr>
<th>Report ID</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Primary Outcome</td>
<td>None</td>
<td>Comparison of the rates of first attendance to the community-based service</td>
<td>-</td>
<td>Four weeks of smoking cessation</td>
<td>Self-reported Quitline use at 30 and 90 days</td>
<td>6 month 7-day point prevalence</td>
<td>Self-reported, repeated point prevalence of tobacco use at 6 weeks and three months</td>
</tr>
<tr>
<td>Secondary Outcome 1</td>
<td>Intervention time</td>
<td>Comparison of intervention rates at the community-based service at 52 weeks from the intervention period</td>
<td>Telephone contact with the smoking cessation program by intervention group at three months</td>
<td>Proportions (%) of smokers who were offered and accepted behavioural support or pharmacotherapy as inpatients</td>
<td>Self-reported abstinence from cigarettes at 30 and 90 days</td>
<td>1 month, 3 months and 12 months 7-day point prevalence</td>
<td>Number of quit attempts</td>
</tr>
<tr>
<td>Secondary Outcome 2</td>
<td>Treatment enrollment (attending at least one session)</td>
<td>A comparison of attendance rates at the community-based service at 55 weeks among the three groups while reattending the hospital-based program</td>
<td>Patient attendance at the smoking cessation program by the intervention group</td>
<td>Proportions (%) of smokers who were discharged with cessation therapy and with post-discharge support arranged</td>
<td>-</td>
<td>Sustained abstinence at 3 months, 6 months and 1 year.</td>
<td>Level of readiness to quit in the next 30 days</td>
</tr>
<tr>
<td>Secondary Outcome 3</td>
<td>7-day point prevalence abstinence rates at 3 months</td>
<td>A comparison of quit rates at 55 weeks among the three groups while reattending the hospital-based program</td>
<td>Smoking cessation and nicotine addiction in both groups at three months</td>
<td>Proportions (%) of smokers who were given support from a local stop smoking service after hospital discharge</td>
<td>-</td>
<td>Reduction of smoking at 1,3,6, 12 months</td>
<td>Number of cigarettes smoked daily at three months</td>
</tr>
<tr>
<td>Secondary Outcome 4</td>
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<td>-</td>
<td>-</td>
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</tbody>
</table>

Table 6. Common outcomes

<table>
<thead>
<tr>
<th>Report ID</th>
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<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>-------------------</td>
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<td>------------</td>
<td>------------</td>
<td>------------</td>
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<td>------------</td>
<td>----------------------</td>
</tr>
<tr>
<td>Intervention</td>
<td>Fax referral to cessation service + follow-up calls</td>
<td>Sixty min intervention + three more session + pharmacotherapy + appointment with community-based service + follow-up if did not attend</td>
<td>Standardised, scripted counseling from physician + written and oral referral to cessation service</td>
<td>Brief advice + intensive bedside support + prescription of NRT</td>
<td>Brief advice + description of cessation service + brochure + option of referral</td>
<td>Thirty second warn about 1/2 deaths, advise to quit and refer</td>
<td>Ten-15 minute counseling session + referral to cessation service</td>
</tr>
<tr>
<td>How were smokers identified?</td>
<td>Advanced practice nurse assessed the smoking status of all patients</td>
<td>Hospital staff advised most smokers</td>
<td>Unclear</td>
<td>Researchers ascertained smoking status from admission form or if incomplete, by direct questioning</td>
<td>Unclear</td>
<td>Unclear</td>
<td>All parents/legal guardians of children aged 18 or younger</td>
</tr>
<tr>
<td>Method of cessation support</td>
<td>Attend a session</td>
<td>Attend a session</td>
<td>Attend a session</td>
<td>Attend a session</td>
<td>Quitline</td>
<td>Attend a session</td>
<td>Quitline</td>
</tr>
<tr>
<td>Recruiter/Provider of intervention</td>
<td>Hospital staff</td>
<td>Hospital staff</td>
<td>Research staff/Hospital staff</td>
<td>Researchers</td>
<td>Hospital staff</td>
<td>Hospital staff</td>
<td>Researchers</td>
</tr>
<tr>
<td>Measure</td>
<td>Self report point prevalence</td>
<td>Self-report plus CO reading point prevalence</td>
<td>Self report point prevalence</td>
<td>Self report and CO point prevalence and abstinence</td>
<td>Self reported 7 day point prevalence and abstinence</td>
<td>Self reported 7 day point prevalence and abstinence</td>
<td>Self reported 7 day point prevalence</td>
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<tr>
<td>Nicotine dependence</td>
<td>Fagerstrom Test for Nicotine Dependence (FTND) and cigarettes per day</td>
<td>FTND and pack years</td>
<td>FTND</td>
<td>Not reported</td>
<td>FTND and cigarettes per day</td>
<td>Cigarettes smoked per day</td>
<td>FTND</td>
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<tr>
<td>Four weeks point prevalence</td>
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<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes (6 weeks)</td>
<td></td>
<td></td>
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<tr>
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<td></td>
<td></td>
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<td></td>
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<tr>
<td>Three months point prevalence</td>
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<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Three months abstinence</td>
<td></td>
<td>Yes</td>
<td>Yes</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Six months point prevalence</td>
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</tr>
<tr>
<td>Six months abstinence</td>
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<td>Yes</td>
<td>Yes</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>12 months point prevalence</td>
<td>Yes (55 weeks)</td>
<td></td>
<td>Yes</td>
<td></td>
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<td>12 month abstinence</td>
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<td>Yes</td>
<td>Yes</td>
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<tr>
<td>Patient attendance for cessation support</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patient contacted cessation service via telephone at three months</td>
<td></td>
<td>Yes</td>
<td></td>
<td>Yes</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Risk of bias within studies

Table 2 outlines the findings from applying the Cochrane risk of bias assessment. Murray 2013 scored a high risk of bias for 'allocation concealment' (selection bias) due to wards or clusters being randomised before participants were recruited. Richman 2000 had a high risk of bias for 'incomplete outcome data' (attrition bias) as some eligible participants declined to participate, but no reasons for this were reported. Also, Richman 2000 did not use intention to treat analysis which did not take into account lost to follow-up participants outcomes. Lin 2013 scored a high risk of bias for 'selective outcome reporting' (reporting bias) because an outcome not recorded in the method section was reported in the results section, this may have been to supplement the results. The main reason for an unclear assessment result was due to lack of information in the written study.

The definition of tobacco use varied widely between trials. Murray 2013 included current smokers or those that had smoked within four weeks, Lin 2013 asked the question "Do you smoke cigarettes?" those answering yes were eligible, Warner 2011 included current smokers who had a 100-cigarette lifetime consumption, Andres 2011 included adults who smoked at least one cigarette per day, Mahabee-Gittens 2008 included adults who smoked one cigarette in the previous week, Lewis 2009 did not state their definition of tobacco use but did include patients who had recently tried to quit, and Richman 2008 included smokers who answered yes to the question "Do you smoke now?". All studies used cigarette smokers apart from Mahabee-Gittens 2008 who also included chewing tobacco and snuff users.

Risk of bias was relatively low in the seven studies in this review as can be seen in Figure 1. Random sequence bias (selection bias) was low for all studies and blinding of outcome assessment (detection bias) was all low and unclear. There were three studies that had a high risk of bias in one section, Richman 2000, Lin 2013 and Murray 2013. Richman 2000 had a bias around incomplete outcome data (attrition bias) as they did not report the reasons why some eligible patients declined to participate and they also did not analyse the data on an intention to treat basis. Murray 2013 had a high risk of bias due to allocation concealment (selection bias) as the study was a cluster randomised controlled trial and the clusters were randomised before the patients were recruited. Lin 2013 scored high on the selective outcome reporting (reporting bias) as they reported an outcome in the results section that they did not report on in the methods section. This may be quite significant as it is a measure they used to support the validation of the self-reported seven day point prevalence abstinence.

Results of individual studies

Table 7. Results of individual studies using common outcomes measured

<table>
<thead>
<tr>
<th>ID</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intervention</td>
<td>Fax referral to cessation service + follow-up calls</td>
<td>Sixty min intervention + three more sessions + pharmacotherapy + appointment with community-based service + follow-up if did not attend</td>
<td>Standardised, scripted counseling from physician + written and oral referral to cessation service</td>
<td>Brief advice + intensive bedside support + prescription of NRT</td>
<td>Brief advice + description of cessation service + brochure + option of referral</td>
<td>Thirty second warm about 1/2 deaths, advise to quit and refer</td>
<td>Ten-15 minute counseling session + referral to cessation service</td>
</tr>
</tbody>
</table>

200
<table>
<thead>
<tr>
<th>Power</th>
<th>Achieved sample size needed for 80% to detect absolute difference</th>
<th>Achieved sample size needed for 80% to detect absolute difference</th>
<th>Not reported</th>
<th>Did not achieve sample size needed for 80% to detect absolute difference</th>
<th>Achieved sample size needed for 90% to detect absolute difference</th>
<th>Did not achieve sample size needed for 80% to detect absolute difference</th>
<th>Not reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>Four week point prevalence - significant difference between intervention &amp; control?</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Intervention n/N</td>
<td>98/260</td>
<td>-</td>
<td>-</td>
<td>30/149</td>
<td>20/74</td>
<td>16/237</td>
<td>-</td>
</tr>
<tr>
<td>Control n/N</td>
<td>37/224</td>
<td>-</td>
<td>-</td>
<td>24/151</td>
<td>3/46</td>
<td>4/119</td>
<td>-</td>
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<tr>
<td>p value</td>
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<td>-</td>
<td>-</td>
<td>0.301</td>
<td>-</td>
<td>&lt;0.05</td>
<td>-</td>
</tr>
<tr>
<td>Odds ratio</td>
<td>2.10 (0.96 to 4.61)</td>
<td>-</td>
<td>-</td>
<td>Not reported</td>
<td>4.29 (1.27 to 14.42)</td>
<td>2.08 (0.68 to 6.4)</td>
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</tr>
<tr>
<td>Three month point prevalence - significant difference between intervention &amp; control?</td>
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<td>-</td>
<td>-</td>
<td>No</td>
<td>-</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Intervention n/N</td>
<td>5/111</td>
<td>-</td>
<td>-</td>
<td>6/78</td>
<td>-</td>
<td>27/149</td>
<td>17/71</td>
</tr>
<tr>
<td>Control n/N</td>
<td>8/110</td>
<td>-</td>
<td>-</td>
<td>5/74</td>
<td>-</td>
<td>26/151</td>
<td>2/45</td>
</tr>
<tr>
<td>p value</td>
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<td>-</td>
<td>1.00</td>
<td>-</td>
<td>0.896</td>
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</tr>
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<td>Odds ratio</td>
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<td>-</td>
<td>-</td>
<td>Not reported</td>
<td>Not reported</td>
<td>Not reported</td>
<td>3.28 (0.94 to 11.41)</td>
</tr>
<tr>
<td>Six months point prevalence - significant difference between intervention &amp; control?</td>
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<td>-</td>
<td>-</td>
<td>No</td>
<td>-</td>
<td>No</td>
<td>-</td>
</tr>
<tr>
<td>Intervention n/N</td>
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<td>-</td>
<td>-</td>
<td>47/250</td>
<td>-</td>
<td>16/67</td>
<td>-</td>
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<tr>
<td>Control n/N</td>
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<td>-</td>
<td>-</td>
<td>19/219</td>
<td>-</td>
<td>3/43</td>
<td>-</td>
</tr>
<tr>
<td>p value</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>0.06</td>
<td>-</td>
<td>0.05</td>
<td>-</td>
</tr>
<tr>
<td>Odds ratio</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>2.10 (0.96 to 4.61)</td>
<td>-</td>
<td>3.43 (1.00 to 11.77)</td>
<td>-</td>
</tr>
<tr>
<td>Twelve months point prevalence - significant difference between intervention &amp; control?</td>
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<td>No</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>No</td>
<td>-</td>
</tr>
<tr>
<td>Intervention n/N</td>
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<td>22/132 &amp; 26/132</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>14/62</td>
<td>-</td>
</tr>
<tr>
<td>Control n/N</td>
<td>-</td>
<td>26/129</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>3/41</td>
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<td>Not reported</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>3.28 (0.94 to 11.41)</td>
<td>-</td>
</tr>
<tr>
<td>Participants attendance at cessation support &amp; significant difference between intervention &amp; control?</td>
<td>Yes</td>
<td>Yes</td>
<td>No*</td>
<td>Yes</td>
<td>-</td>
<td>No*</td>
<td>-</td>
</tr>
<tr>
<td>-------------------------------------------------</td>
<td>-----</td>
<td>-----</td>
<td>-----</td>
<td>-----</td>
<td>----</td>
<td>-----</td>
<td>----</td>
</tr>
<tr>
<td>Intervention n/N</td>
<td>15/111 (13%)</td>
<td>30/129 (23%)</td>
<td>0/78</td>
<td>80/262 (30%)</td>
<td>-</td>
<td>1/74</td>
<td>-</td>
</tr>
<tr>
<td>Control n/N</td>
<td>1/110 (3%)</td>
<td>9/132 (7%) &amp; 5/132</td>
<td>0/74</td>
<td>21/229 (9%)</td>
<td>-</td>
<td>0/46</td>
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</tr>
<tr>
<td>P value</td>
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</tr>
<tr>
<td>Odds ratio</td>
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<td>Not reported</td>
<td>Not reported</td>
<td>4.22</td>
<td>(2.27 to 7.83)</td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th>Patient contacted cessation service by phone at three months &amp; significant difference between intervention &amp; control?</th>
<th>-</th>
<th>-</th>
<th>No*</th>
<th>-</th>
<th>Yes</th>
<th>-</th>
<th>-</th>
</tr>
</thead>
<tbody>
<tr>
<td>p value</td>
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<td>-</td>
<td>Not reported</td>
<td>-</td>
<td>&lt;0.0001</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Odds ratio</td>
<td>-</td>
<td>-</td>
<td>Not reported</td>
<td>-</td>
<td>Not reported</td>
<td>-</td>
<td>-</td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th>Reduction in smoking at three months &amp; significant difference between intervention &amp; control?</th>
<th>-</th>
<th>-</th>
<th>-</th>
<th>-</th>
<th>-</th>
<th>No</th>
<th>Yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>p value</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>0.06</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Odds ratio</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>0.26</td>
<td>(0.75 to 2.95)</td>
</tr>
</tbody>
</table>

* Zero or very low attendance, so no further analysis performed

Synthesis of results

Due to the variability of participants, interventions, reported outcomes and missing data we decided to conduct a narrative synthesis rather than a meta-analysis. To aid the narrative synthesis we recorded all the outcomes reported by all seven studies in Table 5. We then recorded all outcomes of interest to the review that two or more studies reported on in Table 6. Table 7 reports the results of each outcome.

Narrative Synthesis

Preliminary Synthesis

The most commonly reported outcome among the seven studies was ‘participant attendance at a smoking cessation support session’ which five studies measured. This outcome was very important in answering the review question as it measures uptake. Three studies reported a significant difference in attendance for support between the intervention and control/usual care group. Those studies were Anders 2011 who looked at a simple fax referral to the cessation service who followed participants up compared to the control group who received self-help materials and contact information for the cessation service, Lewis 2009 who delivered a 60 minute session followed by three weekly sessions, prescribed pharmacotherapy and then passed over to the community-based programme with an appointment time compared to 20 brief
intervention and advice to call community-based programme, and Murray 2013 who gave brief advice with intensive bedside support and pharmacotherapy compared to usual care which was generally advice to stop and the offer of support if the patient wanted it. Lewis 2009 and Murray 2013 had the most significant differences between the intervention and control groups and the highest percentage of intervention participants going on to attend the cessation support. The similarities between these two interventions was in their intensity. Unfortunately, Anders 2011 and Lewis 2009 did not report four week point prevalence data, but Murray 2013 showed an almost significant difference (p=0.06) between intervention and control. Looking at the control condition in the Murray 2013 study does show that 'advice to quit and offers of cessation support were then given to patients at the discretion of and in accordance with the usual practice of doctors and health care professionals involved in their care' (p.2).

The seven studies reported on 7-day point prevalence and abstinence from smoking at a range of time points, from discharge to 55 weeks after the intervention. Only one study found a significant difference between the intervention and control 7-day point prevalence rates at four weeks and three months and that was Lin 2013. In Lin 2013 they used physicians who gave a 30 second long advice session warning participants that half of smokers die from a smoking related disease, that they advise them to quit and then referred them to the cessation service. This was compared to the control group who had no intervention which was usual practice at that hospital.

Exploring relationships
Within studies
Two of the studies that produced significant attendance at cessation support results both had high intensity interventions and more than one intervention session, this was also due their participants being inpatients. Five of the studies used hospital staff to deliver the intervention, only Murray 2013 and Mahabee-Gittens 2008 used researchers. This is extremely positive as it reflects real world care (Rigotti et al. 2012). Although Richman 2000 used researchers to identify and recruit smokers to the study. Also only Warner 2011 and Mahabee-Gittens 2008 used a 'Quitline' for cessation support, the other five studies used face-to-face hospital or community sessions. It was these two studies that used a quit line that showed no significant results for quitting. Five of the studies used the Fagastrom Test for Nicotine Dependence (FTND) to measure nicotine dependence (Anders 2011, Lewis 2009, Richman 2000, Warner 2011 and Mahabee-Gittens 2008). Murray 2013 did not report on the measures they used for dependance and Lin 2013 used cigarettes smoked per day. The FTND is a widely used and valid measure of dependence (Heatherton, 1991). All studies used self-report measures of point prevalence or abstinence and self report measures are found to be as reliable as other methods of cessation reporting.

Between studies
There was considerable heterogeneity in the seven studies, which made comparability a challenge for the reviewers. Three studies focused their recruitment on emergency departments (ED) (Anders 2011, Richman 2000 & Mahabee-Gittens 2008), one recruited participants from pre-operative care who were waiting for elective surgery (Warner 2011) and Lewis 2009, Murray 2013 and Lin 2013 recruited participants from a large hospital setting. The setting maybe of significance to the findings as patients in ED may be quite different in characteristics to those studies using in-patients, so not easily comparable. The intensity of intervention varies between studies from 60 minutes for the first intervention in Lewis 2009 to 30 seconds in Lin 2013. According to Rigotti et al, 2012, a low intensity smoking cessation intervention is less than 10 minutes and a high intensity intervention is more than 10 minutes. Out of the seven studies in this review four are of low intensity and 3 are of high intensity. Two of the high intensity ones have produced the best results for attendance at cessation support sessions (Lewis 2009 and Murray 2013). The only study to produce significant results for four week and three month point prevalence was the study with the lowest intensity intervention with a one-off, 30 second
intervention (Lin 2013). Anders 2011 and Lewis 2009 were the only studies that used hospital staff to assess all patients used for the study setting.

Richman 2000 was the only study out of the seven that produce no significant differences between the intervention and control group. The reviewers concluded that this may be due a significant difference of this study and the other six; which was the information physicians gave to participant about the stop smoking programme cost of $200. No other study reported a cost to the participants for attending smoking cessation support.

Narrative synthesis by subgroup
To investigate the relationship between the seven studies further we grouped them into settings and intensity of intervention. The first grouping was exploring the findings of the studies conducted in ED (see table 8). Comparing Anders 2011, Richman 2000 and Mahabee-Gittens 2008 it would suggest that Anders 2011 is the most robust in terms of bias and quality, although the study scored one weak rating in quality this was due to a low proportion of participants completing the study. However, they did record the number of drop-outs and the reasons why. Also Anders 2011 was the only ED study to significantly increase cessation support attendance with their fax referral intervention. Mahabee-Gittens 2008 did report a significant reduction in smoking at three months, unfortunately this was not recorded by the other two studies. The difficulty in comparing the three ED department studies comes also when looking at the participant group as Mahabee-Gittens 2008 looked at parents of children admitted to ED rather than the patients themselves.

Table 8 Studies grouped by setting - emergence department (ED)

<table>
<thead>
<tr>
<th>Report ID</th>
<th>1</th>
<th>3</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intervention</td>
<td>Fax referral to cessation service + follow-up calls</td>
<td>Standardised, scripted counseling from physician + written and oral referral to cessation service</td>
<td>Ten-15 minute counseling session + referral to cessation service</td>
</tr>
<tr>
<td>Method of cessation support</td>
<td>Attend a session</td>
<td>Attend a session</td>
<td>Quitline</td>
</tr>
<tr>
<td>Provider of intervention</td>
<td>Hospital staff</td>
<td>Hospital staff</td>
<td>Researchers</td>
</tr>
<tr>
<td>Power</td>
<td>Sample size achieved to detect difference (80%)</td>
<td>Not reported</td>
<td>Not reported</td>
</tr>
<tr>
<td>% low risk of bias</td>
<td>100%</td>
<td>57% (Had one high risk of bias)</td>
<td>71%</td>
</tr>
<tr>
<td>Quality Assessment</td>
<td>Medium (one weak score)</td>
<td>Medium</td>
<td>Medium (one weak score)</td>
</tr>
<tr>
<td>Four week point prevalence</td>
<td>-</td>
<td>-</td>
<td>No significant difference</td>
</tr>
<tr>
<td>Three month point prevalence</td>
<td>No significant difference</td>
<td>No significant difference</td>
<td>No significant difference</td>
</tr>
<tr>
<td>Six month point prevalence</td>
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<td>Twelve month point prevalence</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Attendance at cessation session</td>
<td>Significant difference found</td>
<td>No significant difference</td>
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</tr>
<tr>
<td>Contact to Quitline made</td>
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<td>-</td>
<td>No significant difference</td>
</tr>
<tr>
<td>Reduction of smoking at three months</td>
<td>-</td>
<td>-</td>
<td>Significant difference found</td>
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</tbody>
</table>
The second subgroup consisted of the other four studies which looked at participants in a more general hospital setting (Table 9.). Even in grouping the remaining four studies we still have quite a difference in setting as Warner 2011 recruited participants waiting for elective surgery and Lin 2013 only recruited male outpatients. So in comparing Lewis 2009 and Murray 2013 we find that Lewis 2009 is a robust study with low risk of bias throughout and a medium rating of quality with one weak score for reliability and validity of data collection tools. Also, as a more robust study we can then look to the intervention, which resulted in significantly more participants attending cessation support. Murray 2013 did however did have a risk of a Type II error occurring, so it was possible the intervention did make a difference to the point prevalence rates, but the sample size did not allow for detection of this.

Table 9. Studies grouped by setting - general hospital setting

<table>
<thead>
<tr>
<th>Report ID</th>
<th>2</th>
<th>4</th>
<th>5</th>
<th>6</th>
</tr>
</thead>
<tbody>
<tr>
<td>Author</td>
<td>Lewis 2009</td>
<td>Murray 2013</td>
<td>Warner 2011</td>
<td>Lin 2013</td>
</tr>
<tr>
<td>Intervention</td>
<td>Sixty min intervention + three more session + pharmacotherapy + appointment with community-based service + follow-up if did not attend</td>
<td>Brief advice + intensive bedside support + prescription of NRT</td>
<td>Brief advice + description of cessation service + brochure + option of referral</td>
<td>Thirty second warn about 1/2 deaths, advise to quit and refer</td>
</tr>
<tr>
<td>Method of cessation support</td>
<td>Attend a session</td>
<td>Attend a session</td>
<td>Quitline</td>
<td>Attend a session</td>
</tr>
<tr>
<td>Provider of intervention</td>
<td>Hospital staff</td>
<td>Researchers</td>
<td>Hospital staff</td>
<td>Hospital staff</td>
</tr>
<tr>
<td>Power</td>
<td>Sample size achieved to detect difference (80%)</td>
<td>Risk of Type II error</td>
<td>Sample size achieved to detect difference (90%)</td>
<td>Risk of Type II error</td>
</tr>
<tr>
<td>% low risk of bias</td>
<td>100%</td>
<td>86% (one high score)</td>
<td>100%</td>
<td>86% (one high score)</td>
</tr>
<tr>
<td>Quality Assessment</td>
<td>Medium (one weak score)</td>
<td>Medium (one weak score)</td>
<td>Medium (one weak score)</td>
<td>Medium (one weak score)</td>
</tr>
<tr>
<td>Four week point prevalence</td>
<td>-</td>
<td>No significant difference</td>
<td>No significant difference</td>
<td>Significant difference found</td>
</tr>
<tr>
<td>Three month point prevalence</td>
<td>-</td>
<td>-</td>
<td>No significant difference</td>
<td>Significant difference found</td>
</tr>
<tr>
<td>Six month point prevalence</td>
<td>-</td>
<td>-</td>
<td>No significant difference</td>
<td>-</td>
</tr>
<tr>
<td>Twelve month point prevalence</td>
<td>No significant difference</td>
<td>-</td>
<td>-</td>
<td>No significant difference</td>
</tr>
<tr>
<td>Attendance at cessation session</td>
<td>Significant difference found</td>
<td>Significant difference found</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Contact to Quitline made</td>
<td>-</td>
<td>-</td>
<td>Significant difference found</td>
<td>-</td>
</tr>
<tr>
<td>Reduction of smoking at three months</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>No significant difference</td>
</tr>
</tbody>
</table>

We also divided the studies by intervention intensity; Rigotti et al. (2012) defined high intensity smoking cessation interventions as more than 10 minutes in duration. Table 10 shows the low intensity interventions.

Table 10. Studies grouped by intervention intensity - low intensity(<10 mins)

<table>
<thead>
<tr>
<th>Report ID</th>
<th>1</th>
<th>3</th>
<th>5</th>
<th>6</th>
</tr>
</thead>
</table>
Table 2 shows that the most robust studies in terms of bias were Anders 2011 and Warner 2011 and both found that their interventions resulted in significantly more participants accessed cessation support. However, Warner 2011 used self-report to record access to the Quitline, whereas Anders 2011 used a more valid measure of attendance at a session recorded by the cessation team. Lin 2013 did report a significant difference for four week and three month point prevalence for smoking cessation and had the lowest intensity intervention out of all the studies. However, there may be difficulties in generalising the findings from this study due to the participants being male outpatients only and the population and societal differences in China compared to the UK and USA.

Table 11. Studies grouped by intervention intensity - high intensity(>10 mins)

<table>
<thead>
<tr>
<th>Report ID</th>
<th>Author</th>
<th>Intervention</th>
<th>Method of cessation support</th>
<th>Provider of intervention</th>
<th>Power</th>
<th>% low risk of bias</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Attend a session</td>
<td>Hospital staff</td>
<td>Sample size achieved to detect difference (80%)</td>
<td>100%</td>
</tr>
<tr>
<td>2</td>
<td>Lewis 2009</td>
<td>Sixty min intervention + three more session + pharmacotherapy + appointment with community-based service + follow-up if did not attend</td>
<td>Attend a session</td>
<td>Researchers</td>
<td>Risk of Type II error</td>
<td>86% (one high score)</td>
</tr>
<tr>
<td>4</td>
<td>Murray 2013</td>
<td>Brief advice + intensive bedside support + prescription of NRT</td>
<td>Attend a session</td>
<td>Researchers</td>
<td>Risk of Type II error</td>
<td>71%</td>
</tr>
<tr>
<td>7</td>
<td>Mahabee-Gittens 2008</td>
<td>Ten-15 minute counseling session + referral to cessation service</td>
<td>Attend a session</td>
<td>Hospital staff</td>
<td>Hospital staff</td>
<td>100%</td>
</tr>
</tbody>
</table>
Table 11 shows the three studies classed as delivering high intensity interventions. Lewis 2009 was robust in terms of low bias and good quality and the intervention also reported a significant increase in attendance to the cessation service.

Discussion

Summary of evidence
In this review we aimed to find out how best to increase the uptake of smoking cessation support in the secondary care setting and we found seven studies which met the inclusion/exclusion criteria. The seven studies varied greatly on aspects like intervention, participants, settings and outcome measures so the results and characteristics of these studies were reported using narrative synthesis. Because we could not combine the results in a meta-analysis the conclusions drawn from the synthesised data was limited. The results of our synthesis did show that overall fax referrals to community smoking cessation clinics and follow-up phone calls do result in a greater uptake of smoking cessation support.

We grouped our studies into settings (ED or general hospital) and by intensity of intervention (low = <10 mins) and found that two studies were more robust than the others in terms of bias and quality; they were Anders 2011 and Lewis 2009. These two studies also looked specifically at uptake of smoking cessation support as they reported on numbers attending smoking cessation sessions and found a significant difference between intervention and control groups. There were distinct differences between these studies as Anders 2011 recruited participants from non urgent ED and used a low intensity intervention and Lewis 2009 recruited participants from the hospital setting in general and used a high intensity intervention. Both these studies used fax referrals and follow-up from the smoking cessation support service.

There was considerable heterogeneity within the studies so it was not possible to identify the key effective components of each intervention. However, these results remain relevant to key groups within the health care system. The 30 second intervention by Lin 2013 is of great interest to clinical care leads as they found significant differences between intervention and control participants 4 week and three month point prevalence. This intervention was the least intensive and carried out by hospital staff, therefore it is applicable the hospital setting and extremely low in cost and implementation time. There were draw backs to be considered in the Lin 2013 study which were the sample they used was male only outpatients and also the hospital was a ‘grade A’, which is not comparable to community hospital health care. Most of the interventions involved the hospital staff or researcher making the referral themselves compared it to usual care or control whereby the participant was either given the contact details of the service or just given information on stopping smoking. This is of importance to commissioners, providers and all
hospital staff to know that giving patients information and contact details is more often than not just not enough to stimulate a move towards uptake of support by the patient.

Limitations
Outcome level limitations
It was the reviewers aim to find studies that also reported how smokers were identified within the secondary care setting. Three of the studies (Richman 2000, Warner 2011 and Lin 2013) did not clearly report how smokers were identified and two studies (Murray 2013 and Mahabee-Gittens 2008) had researchers identify smokers, which is not sustainable once the study period is over. Some of the outcome definitions varied across studies; for example, measure of abstinence or point prevalence was measured by self-report in some and expired carbon monoxide in others, and some used both. Some studies used time point that were not standardised for reporting of smoking abstinence as Mahabee-Gittens 2008 reported at six weeks instead of the usual four. Also Murray did not report the participants’ nicotine at base-line and Lin 2013 did not use the standard measure of nicotine dependence, the FTND. It may be advantageous for studies to give an approximation of the cost of their interventions as this would help inform viability of implementation to secondary care stakeholders.

Study level limitations
Warner 2011 was the only study who measured the constancy of their intervention; the remaining studies did not have sufficient measures in place to make sure the intervention they were delivering was standardised. There may be some issues of generalisability as Lin 2013 only studied male outpatients, Warner only studies patients in for elective surgery, Murray 2013 and Mahabee-Gittens 2008 used researchers to carry out the intervention rather than hospital staff. Other limitations included Richman 2000 did not use any intention to treat analysis, Richman 2009 and Mahabee-Gittens 2008 not reporting sample size needed to increase sensitivity. There may have been some selective reporting as Lin 2013 reported an outcome in the results that was not reported in the methods section, this may have been to bolster the validity of their four week and 3 month point prevalence data. Also the results of these studies may not be entirely generalisable to all hospital patients as most studies excluded patients that were too ill at the time of recruitment, suffering from drug and alcohol dependency or patients with no fixed address or telephone number. These exclusions may make it harder to apply in practice, change effects of interventions and not be applicable to all socio-economic groups.

Review level limitations
The review was restricted to studies reported in the English language only, so it is possible for some studies to have been missed. The review could have included the results of another study not yet published, however, the authors of this study did not respond when contacted (Richter et al, 2012). The exclusion of this study could have had an impact on the overall conclusions of this review. Risk of bias could have been reduced further if both reviewers had conducted the data extraction process and compared results. However, due to time and resources this was not practical.

Conclusions
In this review investigating ways to engage secondary care patients with the stop smoking service we have concluded that even a very brief 30 second intervention can be effective in producing four week point prevalence in smoking cessation. The most robust and efficacious studies used hospital staff to refer patients themselves to the cessation service and for that service to follow patients up after discharge. These results are of particular importance to clinic care leads, commissioners and providers. These conclusions can also be applied to other secondary care settings such as mental health, maternity and rehabilitation as the review by Rigotti et al. (2012) concluded that support was found to be effective regardless of diagnosis. It is with caution that the reviewers state these main findings due to the study and review limitations mentioned above.
The NICE guidance (2013) highlights the need for more research into identifying smokers in secondary care and this review supports that finding as three studies failed to report on how this was achieved. Also, future research should include details of cost to implement interventions as this would aid in the decision making process for major secondary care stakeholders.

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No funding was received to complete this review as it was required for the completion of a doctorate study programme.

Word count
5, 959

References


Review objectives

To identify the best interventions which increase engagement and uptake of smoking cessation support in the hospital setting.

Participants

Patients and visitors in secondary care (inc. outpatient settings, emergency dept., 18+, can include staff, but not exclusively staff) (exc. mental health, pregnancy, walk-in centres and primary care).

Interventions

Identification of smoker, recruitment into stop smoking program, engagement around smoking cessation, very brief advice, staff attitudes, patient attitudes, provision of NRT, provision of support (exc. smoking cessation interventions, pharmacotherapy, studies including other substance misuse).

Outcomes

Referral rates, engagement in local smoking cessation services, use of NRT.

Comparison

Routine care or a control intervention.

Study design

Randomised controlled trial
1 exp smoking cessation program/ or exp smoking/ or exp smoking cessation/ or exp nicotine/
2 limit 1 to (human and english language)
3 (Tobacco or Quit* or Smok* or Stop or Cess* or Giv*).mp. [mp=title, abstract, subject
   headings, heading word, drug trade name, original title, device manufacturer, drug
   manufacturer, device trade name, keyword]
4 limit 3 to (human and english language)
5 (Engage* or Motivat* or Incentive* or Intention or Interact* or Promot*).mp. [mp=title,
   abstract, subject headings, heading word, drug trade name, original title, device
   manufacturer, drug manufacturer, device trade name, keyword]
6 limit 5 to (human and english language)
7 Recruit*.mp. [mp=title, abstract, subject headings, heading word, drug trade name,
   original title, device manufacturer, drug manufacturer, device trade name, keyword]
8 limit 6 to (human and english language)
9 self-selection.mp.
10 limit 9 to (human and english language)
11 Volunteer.mp. or exp volunteer/
12 limit 11 to (human and english language)
13 (Invit* or Enrol* or Enter* or Entry).mp. [mp=title, abstract, subject headings, heading
   word, drug trade name, original title, device manufacturer, drug manufacturer, device trade
   name, keyword]
14 limit 13 to (human and english language)
15 Patient Participation.mp. or exp patient participation/
16 limit 15 to (human and english language)
17 Uptake.mp. [mp=title, abstract, subject headings, heading word, drug trade name,
   original title, device manufacturer, drug manufacturer, device trade name, keyword]
18 limit 17 to (human and english language)
19 exp patient/ or exp patient participation/ or exp patient referral/ or exp patient selection/
   or exp hospital patient/ or exp patient decision making/ or Patient*.mp.
20 limit 19 to (human and english language)
21 Smoker.mp.
22 limit 21 to (human and english language)
23 Consumer.mp. or exp consumer/
24 limit 23 to (human and english language)
25 Service user.mp.
26 limit 25 to (human and english language)
27 (Customer or Client).mp. [mp=title, abstract, subject headings, heading word, drug trade
   name, original title, device manufacturer, drug manufacturer, device trade name, keyword]
28 limit 27 to (human and english language)
29 exp hospitalization/ or exp hospital patient/ or Inpatient*.mp.
30 limit 29 to (human and english language)
31 exp outpatient care/ or Outpatient*.mp. or exp outpatient/ or exp outpatient
   department/
32 limit 31 to (human and english language)
33 secondary care.mp. or exp secondary health care/
34 limit 33 to (human and english language)
35 exp medical personnel/ or Speciali#ed care.mp. or exp physician/
36 limit 35 to (human and english language)
37 (Acute or Hospital*).mp. [mp=title, abstract, subject headings, heading word, drug trade
name, original title, device manufacturer, drug manufacturer, device trade name, keyword]
38 limit 37 to (human and english language)
39 Intervention.mp. or exp intervention study/
40 limit 39 to (human and english language)
41 randomi#ed controlled trial*.mp. [mp=title, abstract, subject headings, heading word, drug trade name, original title, device manufacturer, drug manufacturer, device trade name, keyword]
42 limit 41 to (human and english language)
43 2 or 48
44 6 or 8 or 10 or 12 or 14 or 16 or 18 or 20
45 22 or 24 or 26 or 28 or 30 or 32
46 34 or 36 or 38
47 40 or 42
48 43 and 44 and 45 and 46 and 47