Parental Views of Antenatal Testing and Termination Following a Diagnosis of Cleft Lip

Abstract

Fetal anomaly screening at 18-21 weeks now routinely includes cleft lip. If detected, the manner in which the diagnosis is communicated can considerably influence prospective parents' decision-making in regard to further testing and termination of pregnancy (TOP). Given the known psychological impact of an antenatal diagnosis of cleft lip on prospective parents, this study aimed to explore the decision-making process from the parents' perspective, in order to evaluate how well current practice is meeting guidelines. Data from 217 parents of children born with cleft lip (with or without cleft palate) were collected using a mixed-methods online survey. Quantitative responses were analysed using descriptive statistics, while qualitative data were analysed using inductive content analysis. The majority of respondents reported feeling upset, offended and/or anxious following the diagnosis, due to unsatisfactory information, insensitive comments, and a perceived lack of empathy from health professionals. Some respondents had felt under pressure to make a decision quickly, with a minority having regretted undergoing amniocentesis, and/or experiencing significant distress at having come close to TOP unnecessarily. The findings provide important insight into the ways in which the option to undergo further testing and/or TOP are perceived by prospective parents in the understudied case of cleft lip. A number of suggestions are made as to how the discussion between parents and health professionals could be improved, to ensure that the information and support needs of families affected by cleft lip are fully addressed.

Keywords: diagnosis; cleft lip; amniocentesis; termination; parent perspective; ultrasound; sonography; pregnancy

Introduction

A cleft lip and/or palate (CL/P) is one of the most common congenital conditions in the world, affecting one in every 600-700 infants each year in the United Kingdom (UK; CRANE, 2016). While the cleft itself is surgically repaired during the child's first year of life, the family are expected to engage in a long-term multidisciplinary treatment pathway. Research has also indicated that CL/P and its ongoing associated treatment can impact upon the child's social, emotional and cognitive development (Stock & Feragen, 2016), in addition to parental wellbeing and family functioning (Nelson, Glenny, Kirk & Caress, 2012).

In England, cleft lip was included in routine fetal anomaly screening at 18-21 weeks in April 2010 (Public Health England, 2013), and guidelines were subsequently set out by the Royal College of Obstetricians and Gynaecologists (RCOG, 2010). These guidelines state that parents should be provided with information about the purpose and potential outcomes of antenatal screening in relation to the detection of fetal anomalies before the screening is performed (RCOG, 2010). Should a fetal anomaly be detected, reliable information about the diagnosis and immediate support should be available, and a referral to a doctor with expertise in fetal medicine is recommended (RCOG, 2010). Parents should subsequently be informed that termination of the pregnancy (TOP) is an option prior to the 24-week legal limit. Further testing (in the form of amniocentesis) may be offered to parents in order to determine the likelihood that the cleft is indicative of more serious complications, such as Edwards' (Trisomy 18) or Patau's (Trisomy 13) syndrome. In these cases, TOP may still be offered after the 24-week deadline has passed (RCOG, 2010).

While a diagnosis of cleft lip in their unborn baby may induce feelings of shock, guilt, anxiety and grief among parents (Nelson et al., 2012), the option to undergo invasive antenatal testing may evoke further distress. As well as the risk of miscarriage associated with amniocentesis (Kuller & Laifer, 1995), parents must ultimately make a decision about whether to continue the pregnancy. Previous research in other areas of maternal health has demonstrated that although many women appreciate the option to undergo antenatal testing (Bryant, Hewison & Green, 2005; Inglis, Hippmann & Austin, 2012; Kooij, Tymstra & van de Berg, 2009; Leung, Chay, Chang, Leung, Fung & Lau, 2004), they also lack knowledge regarding testing technologies and possible outcomes (Dahl, Hvidman & Jørgensen, 2011). Equally, few parents are aware of CL/P or its implications prior to diagnosis (Nelson et al., 2012). Interactions with health professionals can therefore have a considerable influence on parents' attitudes toward the diagnosis, antenatal testing, and ultimately TOP (Brajenović-Milić, Babic, Tistic, Vranekovic, Brumini & Kapovic, 2008; Schuth, Karek, Wilhelm & Reisch, 1994).

While a number of studies have demonstrated the potential psychological impact of an antenatal diagnosis of cleft lip on the parents (Nelson et al., 2012; *manuscript under review*), few studies have looked specifically at the process of antenatal testing or TOP following the screening outcome. The aim of the present study was to therefore to explore this decision-making process from the parents' perspective, in order to evaluate how well current practice is meeting the RCOG guidelines.

Methods

Design

Data were collected via an online, mixed-methods survey, advertised to parents of children born with CL/P via the UK-based charity, the Cleft Lip and Palate Association (CLAPA).

Procedure

A mixed-methods survey was designed by CLAPA using the online survey platform, SurveyMonkey. The full survey [Table 1 near here] consisted of nine demographic questions (including the parent's age at the time of diagnosis, and their child's cleft type), 27 quantitative questions (including who provided the diagnosis, and how appropriately respondents felt the option to terminate the pregnancy had been offered), and five qualitative questions (including parents' views of TOP, and the ways in which the amount, timing and content of diagnostic information could be improved).

Institutional approval to analyse the data was provided by (*university*), and a data sharing agreement between the university and CLAPA was agreed and signed by all parties. The study was conducted within the ethical guidelines of the British Psychological Society (2009). Respondents were aware that their contribution to the survey was voluntary, that their data would be kept confidential, that they would not be personally identified during dissemination, and that they could withdraw their data from the study at any time prior to publication.

The survey was advertised on the charity's website, e-newsletters and social media between October 2016 and January 2017. During this time, a total of 1,044 eligible survey responses were collected. The present manuscript describes the quantitative and qualitative data provided by the 217 respondents who received an antenatal diagnosis and were given the option of TOP. Quantitative data were analysed by the first author using descriptive statistics. Since not every respondent answered all of the survey questions, reported percentages were adjusted depending on the number of answers received. Qualitative data were analysed using inductive content analysis. This type of analysis is deemed appropriate when the aim of a study is to succinctly summarise a large body of qualitative data, and when existing theory or research literature on a given phenomenon is limited (Neuendorf, 2017). First, the data were read and re-read, to establish an overall picture of the data. Common themes were then inductively grouped together by the first author in an iterative process (Neuendorf, 2017). A proportion of the data was also coded by the second author to assess reliability. Qualitative codes were then compared, with initial coding reaching an average agreement of 96% (range 94-100%). Any discrepancies were subsequently discussed by both researchers until full agreement was reached. Finally, frequency counts were calculated and exemplar quotes were selected to illustrate each category.

Results

Participant Characteristics

An overview of participant characteristics are provided in [Table 2 near here].

Quantitative data

All survey respondents had received the diagnosis of cleft lip at (*n* = 201/217, 93%) or shortly after (*n* = 16/217, 7%) the antenatal anomaly scan. In the majority of cases, the sonographer had delivered the initial diagnosis (*n* = 183/215, 85%), while in 15 percent of cases (*n* = 32/215), the initial diagnosis had been provided by another health professional, usually another antenatal specialist. Following the diagnosis, 46 percent (*n* = 99/215) of respondents reported that they had been informed about other conditions related to CL/P, such as Stickler's syndrome and 22Q11, while 51 percent of respondents (*n* = 109/215) had reportedly been told about potentially fatal conditions linked to cleft palate, such as Edwards' or Patau's syndrome. A total of 185 respondents had been offered an amniocentesis, of whom 32 percent (n = 70/217) had agreed and 53 percent (115/217) had not agreed to have the procedure. Since all participants were parents of children with CL/P, none of the survey respondents had opted for TOP. Overall, 30 percent of respondents were ‘very satisfied’, and 30 percent were ‘satisfied’ with their diagnostic experience. A further 15% were ‘neutral’, 14% were ‘dissatisfied’, and 11% reported being ‘very dissatisfied’. No differences in overall satisfaction were observed between respondents who received a diagnosis prior to the implementation of the guidelines in 2010 and those who received a diagnosis after 2010.

Antenatal Testing

In addition to these quantitative data, 25 individual qualitative responses relating to amniocentesis were recorded [Table 3 near here]. Participants were not directly asked about antenatal testing, yet these spontaneous responses were considered to be an important aspect of the decision-making process. Three respondents believed that that the option to undergo amniocentesis had been raised appropriately by the health professional, while in contrast, one respondent felt that the option of amniocentesis had been discussed very insensitively. Further, seven respondents reported feeling pressured to have an amniocentesis carried out, and four parents felt that the decision to undergo an amniocentesis had been made for them by a health professional. Two parents reported feeling regretful of the choice to undergo further antenatal testing. Five respondents commented that health professionals had assumed that if a positive result was found on the amniocentesis, then the automatic choice would be to terminate. However, others commented that TOP had only been encouraged as an option due to the identification of an additional condition/syndrome during further testing ($n = 5$). For one respondent, the experience of undergoing the amniocentesis had been an overall positive experience.

Termination of Pregnancy

Of the 217 survey respondents, 73 percent (*n* = 139/191) believed that the option to terminate the pregnancy had been offered neutrally. However, 21 percent of respondents (*n* = 41/191) felt that one option had been encouraged or discouraged over the other, with six percent (*n* = 11/191) stating that they didn't know or couldn't remember. When asked about being offered TOP, respondents reported having a variety of reactions. The most frequent of these was that they had never considered TOP to be an option (*n* = 121/191, 63%). Many respondents reported being greatly upset (*n* = 77/191, 40%) or personally offended (*n* = 53/191, 28%) by the offer of TOP, with a further 28 percent (*n* = 53/191) stating that the offer of TOP was the cause of more worry and anxiety than the diagnosis itself. Ten percent (*n* = 20/191) of respondents reported feeling pressured into making a decision quickly. However, 17 percent of respondents (*n* = 33/191) stated they were reassured to know that TOP was available in the event that antenatal tests were indicative of more serious problems. Five percent of respondents (*n* = 9/191) reported having considered TOP at the point of diagnosis, but later decided against it after receiving more specialist information about CL/P.

In addition to these quantitative data, 181 qualitative responses to being offered TOP were recorded [Table 4 near here]. Thirty-three respondents believed that TOP for a cleft alone should not be offered, and others felt that the very offer of TOP can make the condition sound worse than it is likely to be in reality (*n* = 13). Some respondents felt that the option to terminate had been presented to them neutrally (*n* = 5), whereas others reported that the health professional had delivered the information insensitively (*n* = 5). Nineteen respondents reported that the TOP could be presented alongside misleading information about the condition, which negatively impacted their decision-making. Other respondents emphasised the use of unfavourable language by health professionals (*n* = 4), as well as the offer of TOP being delivered at an inappropriate time (*n* = 26), using inappropriate methods (*n* = 5), and/or without taking the parents' wider context into account (*n* = 10). Seventeen respondents stated that the option to terminate was explicitly encouraged, while for three respondents it was implied to be the preferable option. Three respondents highlighted that the way the offer of TOP had been presented had differed between health professionals. Three fathers also reported feeling excluded from the decision-making process. Pressure to make a decision quickly about whether to end the pregnancy was reported by 12 respondents, and eight respondents stated that they had been offered TOP on multiple occasions, despite having refused previously. Three respondents had come close to opting for TOP; an experience which had later caused them considerable distress. Finally, respondents commented that a difficult diagnostic experience could have a long-term impact on their psychological wellbeing and relationships (*n* = 6).

Discussion

This study is the first to specifically explore parents' experiences of the decision-making process following an antenatal diagnosis of cleft lip since routine screening began in England in 2010. The findings provide important insight into the ways in which the option to undergo amniocentesis and TOP are perceived, and suggest that the RCOG guidelines are not being followed consistently in practice. A number of suggestions are made as to how the discussion between prospective parents and health professionals could be improved, to ensure that the guidelines set out by the RCOG are fully met, and that the information and support needs of families affected by CL/P are effectively addressed.

Of the respondents in this study, the majority had chosen not to have an amniocentesis performed, and due to the advertisement strategy of the survey, none of the respondents had opted for TOP. While some parents felt reassured that these options were available, as has been identified in other fetal medicine studies (Bryant et al., 2005; Inglis et al., 2012; Kooij et al., 2009; Leung et al., 2004), many reported feeling upset or under pressure to decide quickly during conversations with health professionals. Further, some respondents had felt personally offended during the offer of TOP, due to themselves, their partner or another family member having been born with CL/P or a related congenital condition. In this study and in the wider literature, research has acknowledged the use of incorrect terminology, inappropriate comments, and a lack of empathy among health professionals during the diagnosis, as well as an underestimation of the amount of information required to make an informed decision (Aspinall, 2002; Lalor, Devane & Begley, 2007; Schuth et al., 1994). Several respondents in this study also believed they had been given information at the time of diagnosis which shaped their views of amniocentesis and/or TOP, but did not later match their actual experiences of having a child with CL/P.

RCOG guidelines state that all practitioners performing fetal anomaly screening should be trained to impart information, and that they or another health professional should be available to provide immediate support to both parents (RCOG, 2010). Further, that all staff involved should adopt a non-directive, non-judgemental approach, and should not assume, even in the presence of a potentially fatal condition, that the parents will choose to terminate (RCOG, 2010). Prospective parents could therefore benefit from a comprehensible description of the aetiology, likely treatment pathway and prognosis, without health professionals introducing their own values and judgements and/or exceeding their own professional capacity (Aspinall, 2002; Schuth et al., 1994). A referral to cleft specialist clinicians should be made within 24 hours of the first diagnosis, and prospective parents should be directed to reliable lay-led organisations (such as CLAPA) as soon as possible. Such referrals would provide prospective parents with further information, emotional support, and realistic personal accounts of what life is like with a child who was born with CL/P, so that they can decide whether further testing and/or TOP is something they want to pursue. Supplementary written information and trusted website addresses could also be provided (Lalor et al., 2007), particularly given that prospective parents may be emotionally distressed and less able to process complex information in the moment. A minority of respondents in the present study reported having regretted their decision to undergo amniocentesis, and/or had experienced significant distress at having come close to terminating their pregnancy. Specialist information, contacts and resources should therefore be available to prospective parents before a formal decision regarding amniocentesis and/or TOP is required. Equally, both parents should be involved in any consultations, since not only can having their partner present ease women's concerns (Schuth et al., 1994), but fathers/partners may also need information and/or support (Lalor, 2014; Stock & Rumsey, 2015). Neither amniocentesis nor TOP for CL/P (or any fetal anomaly) should be unduly encouraged by health professionals, nor should a refusal to undergo antenatal testing and/or choosing to continue the pregnancy be met with disapproval (Coleman, 2015; Schuth et al., 1994). Ultimately, long-term outcome data relating to children born with fetal anomalies is crucial, in order to provide a more accurate prognosis and better informed prenatal counselling in future (RCOG, 2010).

Taken together, the findings of this and other studies emphasise the importance of providing accurate and unbiased information to prospective parents, in a sensitive manner, so that they can make a truly informed choice (Brajenović-Milić et al., 2008; Lalor et al., 2007; Newberger, 2000). Routine screening for cleft lip was only relatively recently implemented in England (Public Health England, 2013), and the results of the current study suggest that more training, accessible information, and links to trusted specialist organisations may be necessary to ensure that reliable and sensitive information is delivered, and that the RCOG guidelines are followed fully and consistently. Further, the law regarding TOP in the case of a fetal anomaly may be unclear to health professionals (Lotto, Smith & Armstrong, 2017), and as a result, TOP may be offered inconsistently or with varying frequency across units

(Moss, 2001). More research in this area is needed, in order to produce resources to better support health professionals who have the difficult responsibility of communicating the next steps to prospective parents.

A number of limitations of the present study must be acknowledged. Specifically, the survey was only shared with parents who are part of CLAPA's community. While CLAPA's community is considerable, it cannot be assumed that this group, nor the subgroup who responded to the survey, are representative of the CL/P population as a whole. Further, not all survey respondents answered all of the questions, and therefore some data is missing. A mismatch is also evident between the results of the quantitative analysis and the codes identified in the qualitative data, with the qualitative findings painting a potentially more negative picture overall. It is possible that those who recalled a particularly negative experience were more compelled to provide further details of their experiences. It is also possible that some of the answers given by participants were subject to recall bias. All individuals reshape their life stories in an attempt to make meaning out of their experiences (Bailey & Tilley, 2008). In addition, it is known that information can be difficult to assimilate when a person is in distress (e.g. Rodgers et al., 2016). It could be that participants emphasised or diluted certain parts of their experiences; however, this does not make their recalled experiences any less significant. Participants were not explicitly asked to provide further details about antenatal testing (amniocentesis), yet this topic featured often among the qualitative responses, and it was subsequently deemed important to highlight. Future research could explore this aspect of care more directly in order to support or dispute the current findings and to provide further insight. Since all participants were parents of children with CL/P, none of the survey respondents had opted for TOP. It is therefore not possible to gain an understanding of the experiences of those who opted to end their pregnancy on the basis of a cleft lip (with or without associated conditions) from the data presented in this paper.

Nonetheless, this survey provided a large amount of both quantitative and qualitative data on a subject which has to date received very little attention, especially in the context of CL/P. The findings will be used to inform future research in this area, and are pertinent to the ways in which antenatal care is delivered in clinical practice.

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