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ORIGINAL ARTICLE

The impact of participation in genetic research for families with cleft lip with and without cleft palate: a qualitative study

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Abstract Despite being the most common congenital facial anomaly, little is understood about the genetic contribution to isolated clefts of the lip with or without cleft palate (CL/P). 'OzCleft', a family-based genotype/phenotype study, is investigating this further. Participation for families involves various clinical investigations of the child with the cleft, and their unaffected sibling(s) and parents. Informal feedback from individuals involved in OzCleft suggested that participation in this research programme had benefits for families. Taking a qualitative approach, this study sought to investigate this hypothesis further. Semi-structured in-depth interviews were conducted with nine parents who had participated in OzCleft. All parents described participation as a positive experience for themselves and their families. Perceived benefits included a greater appreciation of the cleft treatment experience by unaffected family members. Being involved in a genetic study raised issues for parents regarding the cause of clefting in their child. While some parents found the possibility of a genetic component reassuring, it also raised questions about the potential implications for future generations. Parents were largely unsure about how to communicate this information to their children and the predictive value of this information. This study suggests a lack of genetic understanding and/or perceived value of genetic information by parents of children with CL/P that, in turn, highlights the need for increased support from genetic health professionals in this area.

Keywords Cleft lip with or without cleft palate · Research participation · Genetic information · Family communication

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Introduction

Despite being the most common congenital facial anomaly, little is known about the aetiology of isolated cleft lip with and without cleft palate (CL/P). CL/P is understood to be a multifactorial trait; however, the specific genes and/or environmental exposures that contribute to the condition are unclear (Saikrishna et al. 2011). A growing body of literature suggests that CL/P is not the binary trait as was previously understood, but rather a continuous spectrum with other family members exhibiting subclinical phenotypes, for example, facial asymmetries and dental anomalies (Ferrario et al. 1994; Letra et al. 2007). Therefore, the familial implications of conditions such as CL/P will become increasingly understood as the underlying causative genetic factors influencing the phenotypic spectrum become clearer.

Since 2011, families who have a child with CL/P have been invited to participate in the OzCleft Study which is a genotype/phenotype study based upon the Pittsburgh Orofacial Cleft Study and conducted at the Murdoch Childrens Research Institute (MCRI) in Victoria, Australia (Weinberg et al. 2006). OzCleft aims to contribute to the broader understanding of the causes of CL/P using a genotype/phenotype design. Unaffected and affected family members are being recruited in order to investigate the possibility of subclinical manifestations of the condition.

The OzCleft study is unusual in that family members who are not identified as having the condition undergo similar clinical investigations to those usually reserved for the individual with CL/P. These include a genetic assessment and DNA sample, 3D photographs, dental examination, impressions and radiographs, ultrasounds of the upper lip and a speech assessment. In conducting this study, researchers were increasingly receiving feedback regarding unanticipated benefits of participating. These appeared to include an increased ability or opportunity to discuss CL/P, enhanced empathy for the individual with CL/P, and an insight into the treatment experience. No research exists investigating the effects of participation in such research and this study sought to investigate the impact further.

In accordance with family systems theory, the diagnosis of a condition, such as CL/P, in one family member has repercussions for the whole family (Patterson and Garwick 1994). When considering the overall effect of having a child with CL/P, the majority of the literature is focussed on mothers' experiences, with a particular focus on initial diagnosis and early feeding difficulties. This is despite the fact that the current protocol for CL/P can result in the child, and therefore, the whole family becoming involved in a treatment process that lasts in excess of 20 years (Cadogan et al. 2009).

The way a family communicates in response to a change is critical to managing the demands of a chronic condition, and family discussions regarding CL/P are potentially beneficial (Branstetter et al. 2008; Craft et al. 1985; Slifer et al. 2003). Benefits for the child with CL/P can include reinforcing the presence of features unrelated to CL/P that make the child valued, thereby positively influencing their self-perception (Slifer et al. 2003). For siblings, benefits may include reduced stress as a result of an understanding of what is occurring (Craft et al. 1985). However, while the possible benefits are discussed in the literature, how families communicate and with what frequency and success this occurs in relation to CL/P have received minimal focus.

Communication of genetic information involves informing family members of their health and reproductive risks related to a particular condition present in the family (Forrest et al. 2008b). This communication is often discussed in the literature in terms of barriers and facilitators, including 'cultural, familial and individual factors' (Forrest et al. 2003). Factors affecting the dissemination of genetic information have been found to include existing communication patterns in families, the degree of relatedness, gender, the inheritance pattern and perceived severity of the condition, understanding and clarity of the genetic information and the ability to act on that information (Claes et al. 2003; d'Agincourt-Canning 2001; Hughes et al. 2002; Kenen et al. 2004; Lehmann et al. 2000; Sorenson et al. 2003). Parental communication of information about a genetic condition to their children can be a challenging and distressing process but is optimised when children are informed gradually throughout their childhood (Metcalfe et al. 2011; Rowland and Metcalfe 2013).

The literature regarding communication of genetic information is focussed on single-gene disorders: hereditary cancer and general genetic conditions, due to the obvious implications for at-risk family members (Forrest et al. 2008b; Wiens et al. 2013). No studies were identified that examined family communication of multifactorial conditions such as CL/P where the inheritance pattern is less clear. Nevertheless, there is a growing awareness and interest in genetic counselling for multifactorial conditions, such as psychiatric disorders and diabetes (Austin et al. 2008; Hippman et al. 2013; Peay et al. 2008; Waxler et al. 2012). As this model of multifactorial genetic counselling becomes more commonplace and accepted, it is likely that a greater understanding of the issues experienced when communicating to family members about multifactorial conditions may become more evident and potentially highlight the need for support in this area. Therefore, this study sought to explore families' experiences of participating in the OzCleft Study and how this participation impacted family communication about CL/P.

Methods

Given the exploratory nature of this study, a qualitative methodology was employed. Ethics approval was granted by the Royal Children's Hospital Human Research Ethics Committee (ref no. 32074A), Victoria, Australia, and registered with The University of Melbourne (ref no. 1238057) for the researcher (LJD) to conduct semi-structured interviews with parents who had participated in OzCleft with their families.

Recruitment

Participants were sampled from the families who had participated in the OzCleft Study. At the time of recruitment, 44 families had participated and from these, ten families were randomly selected. The parents of these families were mailed recruitment letters, including participant information and consent forms. The recruitment letter included an opt-out option if parents did not wish to receive any further contact. Follow-up phone calls were conducted after a period of 2 weeks by the OzCleft project manager (SR) if no contact had been made. Potential participants were required to make contact with the researcher (LJD) to arrange an interview.

Interviews

Semi-structured in-depth interviews were conducted by one researcher (LJD). In accordance with the flexible nature of this qualitative research design, an interview guide was used rather

than strict interview questions, giving participants the opportunity to describe particular relevant issues for them in more depth (Liamputtong and Ezzy 2005). The following themes were explored in the interviews: parents' experiences of participating in OzCleft, parents' perceptions of their (unaffected) child/ren's experience of participating in OzCleft, families' communication about CL/P, perception about their child's CL/P treatment and any changes to this after participating in OzCleft.

Parents from the same couple were interviewed separately where possible; however, if it was not possible, or the participants preferred, a joint interview was conducted. While the aim was to interview couples separately to avoid the influence of their partner on descriptions of their experience of participating in OzCleft, joint interviews were made available to maximise potential participant numbers (Hertz 1995; Taylor and de Vocht 2011).

Participants were given the option of face-to-face or telephone interviews that were digitally recorded, transcribed verbatim and de-identified. Data were not analysed for differences between telephone and face-to-face interviews or for differences between couples interviewed together or separately. Pseudonyms were assigned and used throughout the transcripts, and identifying information about the participants was removed. To ensure analytical rigour, the researchers (LJD, LEF, MAS and NK) initially coded the interview data independently into repeated themes and ideas and these emerging themes were compared to reach consensus (Strauss and Corbin 1990). One researcher (LJD) continued an iterative process of reading and rereading the transcripts to thematically analyse the data using a constant comparative approach (Strauss and Corbin 1990). This enabled the identification, comparison and coding of themes within and between interviews (Braun and Clarke 2006).

Results

Eight interviews were conducted with nine participants, consisting of six mothers and three fathers from six different families. Of the three couples who participated, two were interviewed separately and one couple was interviewed together. Three mothers were interviewed individually, and their partners did not participate in the study. The interviews ranged from 20 to 100 min; six were conducted face to face, and two by telephone. Face-to-face interviews were held at a range of locations including participants' homes, place of work or neutral locations, such as cafés.

All families lived in Victoria, Australia and identified themselves as 'Australian'. At the time of interview, all children who had participated in OzCleft were less than 18 years of age. To protect the identity of the participants, family characteristics are not described individually. The characteristics of the probands are detailed in Table 1. All probands had unaffected siblings who participated in OzCleft; four families had one unaffected sibling, and two families had two or more unaffected siblings.

Overall OzCleft experience

All of the parents interviewed described their experience participating in OzCleft as positive, both for themselves and their families, and no negative feedback was received. Participants described their experience as 'great fun' (David) and the project as 'fantastic' (Emma) and 'well organised' (Julie).

Family-based approach

Many participants expressed their appreciation for being able to participate in the study as a whole family and that the unaffected siblings enjoyed taking part and gained an appreciation for their sibling with CL/P experiences relating to treatments.

> Oh it was good! It was nice to be able to do something as a family that was related to the cleft… instead of the focus being on her [proband], it was all of us, together, which was kind of nice. (Melissa)

> the kids liked doing it, they liked doing the tests and thought it was pretty interesting and so it was all quite easy. (Julie)

> there was also that for her [proband] to see us all doing medical sort of things, exams and things, things that she has to go through, I thought might be good, and I thought it might be good for [unaffected sibling] because he doesn't really have anything medical to go

Table 1 Characteristics of the probands whose parents participated in the interviews

	n
No. of probands	6
Sex	
Male	3
Female	3
Time of diagnosis	
Prenatal	4
Postnatal	2
Cleft condition[a]	
Unilateral cleft lip and palate	4
Bilateral cleft lip and palate	1
Unilateral cleft lip only	1

[a] Note all probands have been referred to as having 'CL/P' in this manuscript

through so, and he was really good about it actually, better than what I thought he'd be. (Emma)

Benefits of participation

When initially asked about their experience, some parents indicated that they had not felt the OzCleft participation had any significant impact on their families.

It was just something that we did that day, and then we all moved on…We haven't all dwelled on it or anything like that. (Emma)

We don't talk about the cleft any more or less than we did before the study. (David)

Despite this, many parents then went on to describe benefits for individual family members, including themselves, the child with CL/P, their siblings and their partners. This is one of the advantages of qualitative research in that had the information been sought using quantitative methods, perhaps only the initial responses would have been recorded. Benefits largely related to a perceived appreciation from unaffected family members for what their child with CL/P experiences.

Sitting in the chair and having people poking you and prodding you gives you a bit more of an understanding of what she [proband] does - quite happily!…It just gives you a bit more respect for her. (Melissa)

Yeah I think he [sibling] can appreciate what Nick [proband] goes through… even though Nick's missing school for appointments… it's not a fun thing that he's going through. (Carol)

However, this sense of appreciating the treatment experience of their child was not felt by all.

I don't think we'll ever ever know what Jack [proband] goes through … the tests didn't make me feel any closer to his life. (Christopher)

Participating in a genetic study

Understanding CL/P as a genetic condition

An unanticipated finding in this study was that for some parents, participation in OzCleft led to a change in their understanding, or acknowledgement, of the genetic contribution to the condition CL/P. Some parents described wondering whether they had done something 'wrong', particularly during pregnancy, to cause the CL/P in their child. Participating in OzCleft, a genetic study, led to a change in these beliefs.

I wasn't really aware until that point just how genetically based the condition is … nobody had really made that very clear. (Jane)

The only thing that changed with me was that my theory of the stress [as the cause of CL/P in proband] probably went out the window… they think, it's more genetics. (Carol)

Reactions to CL/P as a genetic condition

The impact of this increased understanding of the genetic contribution to their child's condition varied between participants. While one parent (Melissa) described the focus on genetics as 'reassuring', another (Carol) indicated that this information was upsetting.

I guess it just probably makes…more sense, that's probably the comfort that I would take from the study … it's not just a completely random thing. (Melissa)

It would almost upset me if I found out that it's going to carry on through the family, like the (children) are going to carry that gene and I'd be like… oh I've started that off! (Carol)

Communicating genetic information

In investigating parental attitudes and understanding of the genetic component of CL/P, the communication of this genetic information amongst these families was also explored. On the whole, parents were largely unsure about the relevance and importance of communicating this information to their families. In addition, many indicated that this was not something they had previously considered. One parent described discussing the genetic component of CL/P with their siblings after their child was diagnosed:

there was never any discussion about that [genetic component of CL/P] (Julie)

Intention to communicate

When asked about their intention to communicate genetic information regarding CL/P, many parents were undecided about whether to raise this issue with their children.

To be honest I've not even ever really thought about it!… I suppose you'd probably have a chat about … if they have children, but I guess they're [Julie's children] quite young still. (Julie)

> Probably! Yeah… if they went on to get married and have children, yes you would certainly mention… I suppose?… I don't know! I've never really thought of that! (Carol)

> It is, isn't it [important]? Because it's going to make a difference to their lives… at least they would know… there's this much chance … that you could have a cleft… I don't know. I think life's a bit of a chance isn't it though? … I think you just sort of have to deal with what you get sometimes in life. (Emma)

Approach to communication

Parents were often unsure about how to go about communicating information about CL/P to their family members and children. Participants indicated that they were concerned about causing anxiety to family members and were concerned about the burden this information could cause their children.

> in the email we sent out to family and friends … we did indicate that sometimes clefts just happen, and sometimes … you're genetically dispositioned to have a cleft … just inferred that … that may be the case that it might be genetically linked but let them [Amanda's siblings] … come to that conclusion themselves. We certainly didn't want to plant any ideas in other people's minds or create anxieties for other people. (Amanda)

> I don't even know how you'd tackle that to be honest, I wouldn't even know where to start… I mean it's hard enough … giving them sex education, then you have to throw that on the top of it. That's pretty full on for any kid I would have thought. (Emma)

> If there's no more information than there is available now it's probably not something that we'd openly talk about because … it'll come into his [proband] mind before it comes into ours … it'll be his own inner demon that'll play to him … it's one of those difficult ones… do you sow the seed of doubt into his mind and raise it as a topic, or not? (Christopher)

Despite being unsure about how to approach the discussion, one parent indicated that they felt the best person to have this conversation with the child was the parents themselves.

> It's one of those things… better that it comes from you [parent] than comes from somebody else. Whether that's … a friend who thinks they're being a friend that's done a bit of reading online … which is always a problem these days I think. Kids have far too much knowledge, they Google everything! (Emma)

Discussion

Overall OzCleft experience

This study found that parents valued the family-focussed nature of the OzCleft Study protocol. Participants appreciated that the emphasis was not solely on the child with CL/P, as is usually the case. This was described as leading to an increase in understanding and respect from themselves and their unaffected children. Given the unique nature of the OzCleft Study design, it was not possible to find this same perceived effect in other clinical studies described in the literature. However, with the ever increasing focus on genetic studies, viewing the family as a research unit is not uncommon (Chen et al. 2003). However, as Chen et al. (2003) state in their study into the genetic aetiology of autism, the impact of involving parents and unaffected siblings in clinical research is largely unknown. This study suggests that the participation of both affected and unaffected family members in a clinical genetics study can be a positive experience for all involved.

The importance of integrating siblings into both the treatment process and research has been reported in the cancer literature (Snethen and Broome 2001). Studies suggest that increased education and knowledge about a child's condition can lead to greater empathy in their sibling and understanding when there is an unequal distribution of family resources, such as parental attention (Labay and Walco 2004; Murray 1998; Snethen and Broome 2001). This is in line with some of the reported benefits from this study.

Participation in a genetic study

For many participants, OzCleft's emphasis on the genetic aetiology of CL/P came as a surprise. Participation in itself led to an increased understanding of the genetic aspect of CL/P, and this was met with mixed reactions. As has been described in the literature, many parents had their own theories about what may have caused CL/P in their child, many of which included concerns that it was something that they had done to cause the condition (Beaumont 2006; Cadogan et al. 2009; Kuttenberger et al. 2010; Martin 2005).

These reported feelings of guilt or blame and theories about what may have been done wrong in a pregnancy highlight the importance of clear information, support at the time of diagnosis and in particular reassuring parents that it is not their fault (Beaumont 2006; Cadogan et al. 2009; Colabrese 2010; Kuttenberger et al. 2010; Martin 2005). Parents have also previously reported frustration at a lack of information about the cause and possible explanations for CL/P (Nelson et al. 2012). While a genetic explanation would in some instances remove, or place less focus on the idea of parental wrongdoing during pregnancy, a genetic aetiology is not entirely reassuring. 'Guilt over potentially passing on a "bad gene"

to children is one of the most powerful reactions of people with genetic conditions' (McDaniel 2005).

The reactions of parents to OzCleft's focus on the genetic component of CL/P perhaps suggest that either the genetic nature of CL/P had previously not been made clear or that parents did not understand the genotype/phenotype aim of OzCleft. This raises the possibility that there is a need for health professionals involved in CL/P care to discuss the genetic nature of the condition to families more openly (Weil 2000). This gap in understanding about the genetics of CL/P has been reported previously, with one study finding that the majority of parents incorrectly estimating the recurrence risk for CL/P which suggests the need for more effective education (Colabrese 2010). In addition, participants' lack of understanding about the genotype/phenotype aim of OzCleft raises questions about whether the participants gave informed consent for participation in this study. However, Dixon-Woods et al. (2007) suggest that challenges around gaining informed consent are a 'persistent and incorrigible feature' of research participation. Hence, research teams need to be aware of this 'gap' in understanding as participating families may not understand the genetic component and may take home some inappropriate messages from involvement in a study.

Communication of genetic information

Many parents' responses indicated that they had not previously considered communicating genetic information to their families, which was perhaps linked to their limited understanding, or consideration of CL/P as a genetic condition. Nevertheless, most of the issues raised by participants could be considered barriers to genetic communication. As all the children who participated in OzCleft were under the age of 18 years, reproductive implications were not considered a priority by parents (Aktan-Collan et al. 2011). The notion of linking the disclosure of genetic risk information with life events, such as marriage or starting a family in this study, has been reported elsewhere (Forrest et al. 2003; Metcalfe et al. 2008, 2011). However, a meta-synthesis of research into family communication of genetic information indicates that it is beneficial for children to know of their genetic risks at an earlier age (Metcalfe et al. 2008). This enables them to grow with and adapt to the information, rather than waiting until they are adults which can lead to resentment due to feeling that information has been kept from them (Metcalfe et al. 2008; Plumridge et al. 2011).

Parents' own attitudes and values towards CL/P as a condition also appeared to be intricately linked with their consideration of communicating genetic information. In turn, this attitude to CL/P is interlinked with their attitude towards their own child, the proband. It is unknown from this study to what extent parents' attitudes towards CL/P and its impact on their child, and concerns for future children, are matched by the proband, their siblings and extended family members.

Another issue raised by this study was that parents were unsure about the utility of this genetic information. Given that CL/P is a multifactorial trait, no finite reproductive risk values can be given to family members and individuals with CL/P, which increases the complexity of communicating information about CL/P to family members (Austin et al. 2008). Additionally, there is no prenatal or preconception diagnostic genetic testing available for CL/P as causative genes for the condition have not yet been identified. Instead, prenatal diagnoses of CL/P are made via ultrasound and while some cases of CL/P are identified during the first trimester screening, most are not confirmed until the second trimester screening scan usually offered between 18 and 20 weeks gestation (Berggren et al. 2012; Guyot et al. 2013). This has reproductive implications for parents, individuals with CL/P and their family members, as the only preventative option for CL/P is a late termination of pregnancy. The limited reproductive options together with the lack of clarity about recurrence risks as reported in other CL/P studies and parents indicating that a 'whatever percent chance' was not considered useful are likely to impact communication about CL/P to children and family members (Nusbaum et al. 2008).

Forrest et al. (2003) also found that the level of certainty relating to individuals' understanding of their own risk affects their communication to other family members. It has been suggested that non-genetic (environmental) risk factors 'divert attention away from potential risk to other relatives' and creates uncertainty about what to tell relatives. While this finding was in relation to hereditary breast and ovarian cancer, which is dominantly inherited, the development of CL/P is also believed to be influenced by environmental triggers, and thus, communication of genetic information may be complicated by this same factor.

While some parents indicated that they thought communication was important, and would inform their children about the genetic risk of CL/P, other parents were unclear as to whether they would do the same. The notion of wanting to inform their children themselves rather than have somebody else inform them has been reported elsewhere, with the same meta-synthesis indicating that where open communication did not exist, children were more likely to find information from inaccurate sources (Labay and Walco 2004).

Some parents in this study described planning to wait for their child to bring up issues relating to genetics and future family planning rather than initiating the conversation themselves. However, studies have suggested while parents wait for children to raise the issue of recurrence risks, children also act in a way to protect their parents by not asking them directly about their genetic risks (Metcalfe et al. 2008; Plumridge et al. 2012). This potentially represents an unmet communication need for children.

Uncertainty about how to discuss this information with children and other family members could also be viewed as a barrier. Metcalfe et al. (2008) found that parents felt a lack of support from health professionals in regards to communication of genetic information. This provides an opportunity for intervention by health professionals, particularly genetic counsellors, who are well placed to assist families with communication of genetic information to family members (Wiens et al. 2013). Wiens et al. (2013) recently published family genetic risk communication framework developed through a systematic review and based on the theory of planned behaviour, offers a 'parsimonious explanation of family communication factors to act as a bridge to alterations in clinical practice' (p 239). This framework may be useful to develop and trial an intervention that provides increased support to families with CL/P by genetic counsellors to assist and support families when communicating multifactorial genetic information about CL/P to family members (Wiens et al. 2013). Prior research suggests that increased support from genetic counsellors can increase communication of genetic information in families (Forrest et al. 2008a).

This study does have a number of limitations, particularly the small sample size and the potential bias of the participants who were willing to take part in an interview. As this study was undertaken to fulfil the requirements of the research component of a Master of Genetic Counselling qualification, the student researcher had 1 year to complete the study. Therefore, ten families were invited to participate to ensure the research was completed within the 1-year time frame, resulting in the small sample size. The parents who agreed to participate are possibly more highly motivated to take part in further research in addition to their OzCleft participation, which may have resulted from a positive experience participating in OzCleft. Nevertheless, a dearth of evidence exists regarding families' experiences of participating together in this type of research. Despite the small sample size and potentially highly motivated participants, this study does contribute to understanding of experience of participation and the impact of research participation on families' communication about CL/P. Interestingly, even highly motivated parents foresee challenges in communicating information about CL/P to their children.

Conclusion

Due to the inherent nature of this small qualitative study, the findings cannot be generalised to all individuals and families with CL/P, or indeed all of the families who participated in OzCleft. Instead, however, the aim has been to describe the experiences of those involved in this research in depth, and provides some important implications for involving genetic health professionals in cleft care. This study suggests that there can be benefits for participation in family-based clinical research, including a greater understanding and empathy for the treatment experience of the child with CL/P. Parents' lack of understanding of the genetic aspect of CL/P identified in this study has important implications for wider cleft care; a greater emphasis on the genetic nature of this condition may be required when talking to families about CL/P, both at the time of diagnosis and throughout the treatment experience. Families may also require greater support in disseminating this genetic information amongst their families, which could be facilitated through the integration of genetic health professionals, including genetic counsellors and clinical geneticists, into the CL/P team where this is not already the case.

This study represented the views of parents, both in relation to the OzCleft study and the wider CL/P experience, including their views towards CL/P as a genetic condition, and the value of genetic information about the condition. Further research investigating the views of both siblings and children with CL/P in relation to these matters would be beneficial.

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Compliance with Ethics Guidelines *Conflict of interest* Lynley Donoghue, Margaret Sahhar, Ravi Savarirayan, Supriya Raj, Nicky Kilpatrick and Laura Forrest declare they have no conflict of interest.

Statement on human rights All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

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