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## **Making sense of a ‘de novo’ genetic syndrome: genetic responsibility and the enduring significance of ‘family’**

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### **Abstract**

This article examines the implications for parents and family members when a child is diagnosed with a genetic syndrome. In particular, it describes how practices of understanding are shaped when the syndrome occurs ‘de novo’, that is, when it has not been inherited from either parent and where there is no family history. Despite a significant body of research exploring the social implications of genetic disease and diagnostic technologies, sociological understandings of the implications of a de novo mutation are considerably limited. This article draws on semi-structured interviews conducted with twenty three parents of children diagnosed with 22q11 deletion syndrome, a syndrome associated with high rates of de novo cases. Three themes were identified: ‘lay’ understandings of genetics; making genetic connections and genetic gatekeeping. Overall, this article articulates and confirms the enduring significance of family for contextualising health and illness.

**Key words:** *Genetic syndrome, family communication, responsibility, blame, family history*

## Introduction

This article explores the implications for parents and families when a child is diagnosed with a genetic syndrome caused by a new mutation. Drawing on the accounts of parents of children diagnosed with 22q11 deletion syndrome<sup>1</sup>, three themes were identified: ‘lay’ understandings of genetics; making genetic connections and genetic gatekeeping. The context for this article is the wealth of research exploring the social and ethical implications of developments in genetic knowledge. A significant body of literature within the social sciences focuses on individual *and collective* responses to these developments. At the personal level, a genetic diagnosis or identification of genetic risk can alter an individual’s sense of identity (McConkie-Rosell, *et al.*, 2008). Many authors recognise that in contrast to other types of disease, a genetic syndrome also has specific and significant implications for others (Hallowell *et al.*, 2003).

‘Genetic responsibility’ has emerged as a heuristic device to examine how or whether genetic knowledge shapes social relationships and the obligations one feels towards oneself and others (Hallowell, 1999; Novas & Rose, 2000; Arribas-Ayllon *et al.*, 2008; Weiner, 2010). Genetic technology informs a redefined concept of ‘responsible’ parentage involving understanding risks and making informed decisions in light of genetic knowledge (Lupton, 2003). As Nelkin and Lindee (1995) assert:

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<sup>1</sup> 22q11 deletion syndrome has an interesting history of nosology which is as much about political preference as it is about medical classification (Author XXXX). The parents in this study used a variety of names including 22q11 deletion syndrome, velocardiofacial syndrome and DiGeorge syndrome. To gain a semblance of consistency and avoid confusion, in this article I employ the label 22q11 deletion syndrome throughout.

[Genetic] explanations have a double edge, for while they shift responsibility to DNA, they create a new biological form of blaming – for the ‘flawed’ parents who pass on ‘bad genes’, for those who knowingly take genetic risks (1995 p129)

However, those who are implicated by these new technologies are not just parents or potential parents. The relationships, including wider kin, which are given prominence through these developments are now described as ‘risky’ (Featherstone *et al.*, 2006) and mobilised through a “web of genetic connectedness” (Novas & Rose, 2000). The significance of these relationships are reflected in the construction of risk and responsibility (Walter *et al.*, 2004; Gaff *et al.*, 2007) and in exploring adaptation and familial communication strategies (Forrest *et al.*, 2003; Keenan *et al.*, 2005, Gregory *et al.*, 2007). However within these genetic networks, the obligations and responsibilities that members perform and display towards each other are complex. Although genetic diagnosis might bring new explanations of disease causality, the context within a diagnosis, or illness, is given meaning can be characterised through continuity. The home, family and social relationships often provide the location within which health and experience is experienced. What makes a ‘family’ and how roles and relationships are defined and enacted has been extensively documented. Despite many changes to the structure of the family, many of the obligations and responsibilities that individuals feel towards each other are constant yet continuously being negotiated (Finch & Mason, 1993; Finch, 2007).

When a child is ill or receives a diagnosis, the impact on the family, and on particular members within the family, can be considerable. It is important to note, but possibly not surprising within this context, that genetic technology also has disparate effects. We know that women are often the key communicators of medical knowledge within the family and this role is enhanced by the possibilities of genetic testing (Richards, 1996; Forrest *et al.*,

2003). d'Agincourt-Canning (2001) found for example that women frequently undergo genetic testing if that knowledge will benefit others in the family.

However, the family as the context within which diagnosis and illness are located is important to note for another reason, beyond the provision of care. Families are recognised as key sites where values are shared, and where responses to health and illness are reproduced (Gregory, 2005). This confirms multiple routes for translating and assimilating biomedical information. Parsons and Atkinson's (1992) article 'Lay constructions of genetic risk' marked a decisive moment within medical sociology research. It provided an insight into the experience of those with, or at risk of, a genetic disorder. It highlighted that biomedical concepts such as 'risk' were neither neutral nor universal and thus confirmed that 'lay' perspectives and experiences were valuable sites of knowledge production and sociological enquiry. By documenting how individuals made sense of risk within their everyday experience, the importance of context was established, as described by Michie and Marteau (1996):

New knowledge, especially where science or technology is involved, is assimilated into individual's existing frameworks of knowledge and understanding by anchoring the unfamiliar to what is already familiar, and rendering abstract concepts into something more concrete...knowledge acquisition is therefore a dynamic process that takes place within a social, rather than an individual, context. (1996 p107)

Yet despite this wealth of knowledge about the social implications of genetic knowledge, little is known about how, and whether, families respond to or adjust their patterns of understanding when the condition is not inherited. One significant exception to this is McLaughlin & Clavering (2011). Their anthropological and sociological approach to

understanding the impact on the family following the diagnosis of a genetic syndrome provides new insight, framing the de novo diagnosis as a mark of 'difference' to be negotiated. Specifically they suggest that the identification of a de novo mutation can disrupt a sense of kinship, where the mutation 'distorts the biological bond between parents and child' (p406).

Genetic counselling literature also appears to take the stance that a de novo mutation has substantially different implications. With regard to communication strategies, Leonard and Newton (2010 p202) highlight

“...there are some genetic conditions that are not going to be of relevance to other family members. To this end, it is neither necessary nor desirable that genetics services be involved in disseminating all kinds of genetic information to families. For example, where a genetic condition has arisen de novo in a particular individual it would be inappropriate to undergo family dissemination because that condition would not be present in family members.”

Indeed, the calculation and dissemination of 'risk' information to others in the family might not be appropriate in the case of a syndrome arising de novo. This type of mutation event occurs at the time of fertilisation, which means only future progeny of the child would be 'at risk'. However, this article confirms the multiple ways in which a de novo diagnosis is relevant to parents and relatives. By focusing on the experiences of parents with a child who has been diagnosed with a genetic disease that has arisen through a de novo mutation, or rather in this case, a de novo deletion, this article therefore challenges and extends current understanding. It highlights the importance of recognising similarities between accounts of a genetic disease which is familial and that which is caused by a de novo mutation. In doing so,

it confirms that a genetic disease caused by a de novo mutation has considerable implications for 'family'.

### *What is 22q11 Deletion Syndrome?*

The focus of this study was to explore the meanings of a rare genetic condition, 22q11 Deletion Syndrome. 22q11 Deletion Syndrome is associated with an expansive and variable phenotype of more than 180 potential symptoms, the most commonly reported including congenital heart defects, immune deficiency and cleft palate (Murphy & Scambler, 2005; Shprintzen & Golding-Kushner, 2008). Incidence is estimated to be 1 in 4,000 live births (Scambler, 2000). Diagnosis is either through the clinical presentation of symptoms or molecular genetic testing (for example, using FISH -fluorescence in situ hybridization) which in most cases would identify a deletion of 30-40 genes on chromosome 22. National and international consensus guidelines have been produced as an attempt to create a standardised approach to the complex and multidisciplinary management (Basset et al., 2011; Max Appeal, 2012). Between 90 to 95% cases of 22q11 deletion syndrome are due to a spontaneous de novo deletion (Scambler 2003) which means that there will be no family history of the syndrome in the majority of cases. However, due to the variable nature of the syndrome and the possibility of very mild symptoms, testing for all parents has been recommended, with the provision of support if subsequently diagnosed (Basset et al., 2011). Following their child's diagnosis, parents will sometimes but not necessarily always be offered testing. If neither parent is affected, then siblings will generally not be tested (Shprintzen & Golding-Kushner, 2008). 22q11 Deletion Syndrome is autosomal dominant which means the offspring of an individual diagnosed with the syndrome will have a 50% chance of inheriting the disease.

## Methodology

This paper reports the analysis of twenty three semi-structured interviews conducted with parents of children diagnosed with 22q11 deletion syndrome. The research was part of a multi-site ethnography which also involved interviews with clinicians and observations of conferences and clinical consultations which are described elsewhere (Author XXXX). Ethical approval was gained through the South East Wales Ethics committee and all names have been changed to ensure anonymity.

Fifty families in total were contacted, eight of whom were contacted through a clinical geneticist. The remaining respondents were contacted through a UK based 22q11 deletion syndrome paediatric clinic. Selection was based on a child below the age of 18 who had been diagnosed with 22q11 deletion syndrome. Letters were addressed to ‘The Parents and/or guardian of [child’s name]’. The response rate was 50% and twenty three interviews were conducted by the author. The mother<sup>2</sup> of each child was present during all the interviews. Seven interviews also involved the father and on two occasions the interview involved the mother and grandmother. The age of the child at diagnosis varied considerably, one child was diagnosed prenatally and the oldest was diagnosed at 15 years old. All of the interviews were conducted within the family home except for two which took place outside of the family home at the request of the parents (one in a restaurant and the other in a coffee shop). The interviews lasted between one and two hours.

Semi-structured interviews and a loose interview schedule were employed. These techniques allowed flexibility and were appropriate as the researcher did not have prior knowledge of

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<sup>2</sup> One woman was interviewed as the primary carer of her granddaughter. Where I have included extracts from her interview, I have labelled her as “mother” to preserve anonymity.

individual circumstance before meeting the parent or parents. The topics discussed included the process of diagnosis, current health and future concerns, and ongoing relationships with health professionals. The interviews were audio-recorded and transcribed verbatim by a professional transcription organisation.

The interview transcripts were analysed according to thematic analysis informed by grounded theory. Grounded theory, developed by Glaser and Strauss is a systematic approach to data, allowing themes to emerge as the research progresses (Coffey & Atkinson, 1996; Strauss and Corbin, 1998; Atkinson et al., 2003). Grounded theory provides a research framework that can account for the complexity of social phenomena, by constantly and consistently developing interpretations of data (Glaser, 1992). The transcripts were read multiple times, allowing common themes and ideas to emerge. These were noted, alternative examples found in the texts and comparisons drawn from relevant literature. AtlasTi was primarily used as a management tool to organise the documents and to systemise the coding structure. In the context of the primary aim of the study which was to explore the parental experience of a rare genetic disease, twenty primary codes emerged from the analysis, including communication within the family, communication with health professionals, diagnosis, genetic risk, the role of genetic technology, medical knowledge, sources of support and sources of information.

The significance of individual and collective responses to a *de novo* genetic diagnosis emerged through this analysis. This discrete topic was selected as the focus of this article because of the lack of current research exploring the nature of genetic responsibility in the context of a *de novo* mutation, contrasting with the extensive body of work exploring the social implications of genetic inheritance .

## Analysis

### *Lay understandings of 'genetic'*

A de novo deletion was identified as the cause of the syndrome for the majority of families who took part in this study. This was not the case for two families. One woman who was raising her grandchild suspected that her estranged daughter also had the syndrome. In another case, a woman was tested and received her own diagnosis following the diagnosis of her daughter. Where a de novo mutation had been identified, diverse metaphors were used by respondents to explain the 'new' occurrence of a disease within the family. These highlight a repertoire of explanations based on a continuum between biology and 'bad luck'. Parents drew on the concept of a system error, for example, a 'glitch' [fam02] or an 'anomaly' [fam07]; a random occurrence, 'just one of those things' [fam15] or 'a one off' [fam08] and a biological event, for example, a 'flukey thing' [fam 11], 'a fluke of nature' [fam23] or 'a misplaced piece of chemistry' [fam22]. The descriptions highlight different ways of making sense of a spontaneous mutation, and important for this paper, confirm awareness that there was a biological catalyst that caused the child's illness. However, for many it also raised concerns. In particular, parents reflected on their understanding of 'genetic' and raised questions of blame and responsibility.

Making sense of the de novo mutation led parents to question the assumption that 'genetics' meant 'familial'. One mother explained, "because it is that word genetic you assume that it is from my genes and her dad's genes we make this child and so one assumes that it has come from one of us" [Fam14]. Another mother reported "I wanted to know well if it is genetic how come one of us isn't the carrier?" [Fam23]. The presence of a spontaneous change led

parents to raise the possibility that they played a role in its cause. The role of the mother was particular prominent in parental accounts. The context for this, the biological bond between mother and child, was made overt by one mother, who commented “I understood that it hadn’t come from me but obviously I made her...” [Fam09].

Models of disease inheritance have historically had significant implications for how individuals respond to genetic disease (Henderson & Maguire, 2000). Whereas mendelian routes of transmission emphasise continuity between generations, here parents are required to account for an heritable disease which was not inherited. And here the ambiguity of a de novo mutation is revealed. Whereas the de novo mutation can be understood as ‘a misplaced piece of chemistry’ or ‘just one of those things’, parents can simultaneously hold themselves responsible. Without the possibility of genetic inheritance, a recurring theme in the accounts of parents was concern that something had happened during pregnancy that resulted in the child having health problems. The agency with which parents allocate to their own role in explanations of why a de novo deletion occurred was articulated by one mother “It must have been something I did that made this gene do this” [Fam22]. Attention is focused on the mother and in particular, what she might or might not have done to trigger the event. The following extract represents a typical account of a mother’s search for the cause of her child’s illness.

Mother: When I got the diagnosis as they said you know they said, they tested us all and none of us had it and he said it is just one of those things. It is just, not so much that you want to place the blame but you want to know if there is a reason behind it. So you know it was good in a way to think that I hadn’t done anything, you know you don’t mean, I think that every mother would check that out... it was a real relief to

know there was nothing that we could have done or there was nothing that we had done to cause it because you know both my pregnancies were both planned, the babies, and I took loads of antenatal care and I just couldn't, I mean I wasn't dead young but then I wasn't dead old, I just couldn't work out why and you just think oh if I hadn't had worked, all these things.

[Fam15 Interview with the mother of Henry, diagnosed at six years old]

Reproduction is a key site where the notion of genetic responsibility play out, including making reproductive decisions and how and whether to act on genetic information (Raspberry & Skinner, 2011). Although this mother did not have access to genetic knowledge, she identified and accounted for particular 'risk' factors that might be associated with problems during pregnancy. The 'responsible' mother, as represented in this account, is one who had planned to have children, was an appropriate age for motherhood and took care of herself and her baby during pregnancy. In making sense of the child's illness, pregnancy was constructed as a significant period when the baby's biology was fragile and dependent on the mother's care and attention. The focus on pregnancy here is therefore not surprising, and the rhetoric of motherhood and messages of individual health promotion were vivid within maternal accounts of genetic illness.

A tension is therefore revealed in how families might experience a genetic syndrome which occurs de novo. A syndrome that is genetic yet not familial can focus attention on the activities of the mother during pregnancy. However, the discovery that a syndrome is 'genetic' can absolve parental feelings of guilt, as it confirms that the mother 'had not done anything' during pregnancy. This is a complex cycle of seeking and ruling out cause in which constructions of personal responsibility, and gendered responsibility, play a key role.

### *Making genetic connections*

Some parents in the study were themselves tested following their child's diagnosis and the majority received a negative test. However, the accounts of the parents who received a negative test suggest that there remained a tendency to contextualise their child's diagnosis within their own family history of health and illness. The following account highlights that although parents recognised the syndrome occurred spontaneously, essential questions remained about where the syndrome had come from and who might be implicated.

Mother: the fact that [son] has got heart problems I thought of my dad because my dad had angina...but apparently there is no connection what so ever with that. I don't know much about my own family history because my father is not alive and I don't see my mother and so I don't know about either side of the family in that respect. So I don't know whether there is any history of it in that but there is nothing in me, nothing in [husband]'s family as far as I am aware....it was just a case of it is a fluke of nature, literally.

[Fam23 Interview with the mother of Darren, diagnosed within days of his birth]

In constructing an explanation of the syndrome, parents seek familiarity. In this instance, the mother recognised that the syndrome was due to a 'fluke of nature' yet continued to contextualise this within her family history. Similarities were found between the son's heart condition and the father's angina. The mother identified the barriers encountered, including lack of knowledge about the family and family members. The important point to highlight here is that even when parents have tested negative for the genetic mutation, many persist in

attempting to identify individuals in the family with known or suspected medical problems. These processes reflect established accounts of how individuals and families respond to a genetic diagnosis, involving making sense of symptoms (Featherstone et al., 2006) and assimilating new information within personal stocks of knowledge (Atkinson et al., 2011). The way in which parents attempt to negotiate and attribute responsibility within the family even in the case of a diagnosis linked to a new mutation is significant. The utilisation of family history to make sense of a genetic syndrome caused by a de novo mutation suggests that families draw on a range of resources available to them, including their understandings of the meanings of 'genetic', biomedical knowledge of genetic transmission and their personal and familial experience of health and illness.

Furthermore, the powerful associations of family, in the context of a genetic diagnosis, are such that being able to locate the syndrome within the family can be seen as a positive step. The following extract highlights how kinship can be used as a resource to provide hope for the future.

Mother: Me and George [husband] have been tested for the deletion and we were all right but I still say George has got [the deletion] because to be honest a lot of the things that Jimmy [son] does George does. Like it is hard work for George to put pen to paper and silly little things like if Jimmy is unwell, he is very red under the eyes and George is the same and I do say they have had it wrong with him....

Father: I am a type of guy as well as I like a routine and I like everything in its place, that kind of thing, stupid little things but...

Mother: And silly little things like, where as I would be oh I can do that, George can't do it, and we would always say he has definitely got [it] and I think that he should be

tested again because a lot of things like that, what Jimmy [son] can't do George can't do.....

Researcher: So if the test came back that you had it, how would you feel about that?

Father: Oh it wouldn't worry me.

Mother: We looked at it like that we were hoping that one of us would have had it because like I say I am not the brain of Britain and the same as George but either way I looked at it and we have survived.

[Fam25 Interview with the mother and father of Jimmy, diagnosed at five months old]

Although many parents questioned whether they or family members had the syndrome, these were the only ones to discuss this possibility at length. This occasion could be interpreted as the father attempting to repair the 'distorted bond' between himself and the child as a result of the de novo nature of the deletion (see for example McLaughlin & Clavering, 2011).

However, I highlight here the collusion of both mother and father in their accounting of health and illness. This single extract is important because it reveals how possibilities of inheritance can raise powerful emotions about a child and his future. The father's genetic test was doubted because the result contradicted their own observations of shared characteristics between father and son. Judgements of sameness are not about symptoms of illness but about embodied characteristics, about 'silly little things'. The father's characteristics, liking routine, redness under the eyes and avoidance of writing are all transformed as potential indicators.

Featherstone et al., (2006) identified how individuals perform surveillance on themselves and others in the family, looking for signs of the development of illness. This single extract suggests self-surveillance, but also demonstrates a strong desire to translate the signs into symptoms. This extract is particularly revealing because earlier in the interview both mother

and father had expressed fears for the child's future, that he might not be able to maintain friendships, hold down a job or live independently. This finding therefore might reflect the difficulties of living with uncertainty (Lipinski et al., 2006). Seeking similarities between son and parent becomes a mechanism for predicting the future trajectory of the child's illness and regaining control. Significantly, the desire to repeat the genetic test for this father was not fuelled by concern about the father's health status but it was embraced because it offered hope and potential reassurance. This finding has parallels with McAllister et al.'s (2007) conclusion that in the context of a child's genetic syndrome, parents were not necessarily concerned about their own health.

This example, of hope being found in the father's potential diagnosis challenges our understanding of illness as biographical disruption and suggests how it can become normalised through kinship. In the majority of the interviews parents engaged in normalisation practices: the syndrome was not always cast as problematic nor met with fear or anxiety, reflecting the development of experiential knowledge and familiarity with illness over time (Charmaz, 1991). A family history of disease therefore provides the context through which diagnosis and the experience of illness can be located (Featherstone et al., 2006). The fact that a negative result does not signal the end of the parental quest to understand the source of the syndrome is a key finding of the study, and particularly in the context of parental testing, contributes to a greater understanding of why negative results can fail to reassure (Michie et al., 2003).

### **Genetic gatekeeping**

The discussion thus far has highlighted the complex network of negotiations and meaning making that might surround a genetic diagnosis. Whereas the possibility of genetic inheritance might offer hope for some, parental narratives suggest that the marrying of ‘genetic’ with ‘family’ can also be experienced as a source of conflict that requires management. The following extract identifies how within this context, a de novo deletion can offer protection from blame.

Mother: But Steve [husband] did find [diagnosis] very difficult to cope with ... and I know it sounds daft, but when we had the blood test and neither of us had it so it was literally just a mix in the gene pool, because you didn’t feel you were responsible it was easier to accept in some ways.

Grandmother: He was very worried that it might have been in his side of the family and it worried him.

Mother: I know it sounds funny I was almost wishing it was me because then it took the weight off his shoulders, as it was it was nobody.

[Fam18 Interview with the mother and grandmother of George, diagnosed at eight years old]

This mother not only suggests that she would be willing to accept that she might be implicated as the source or the reason for her child’s illness but also appears willing to take on the burden of cause from her husband. The words of the mother provide further information about accounting for responsibility. As explored previously, the responsible matriarch is one who prepares for motherhood and makes responsible choices. Here however, responsibility is also demonstrated by openness to the possibility of being identified as the cause of her child’s problems. In their accounts, mothers in particular, presented themselves

as ‘open minded’ to the possibility that they played a role in the transmission of illness to their child in terms of both agency (for example, behaviour during pregnancy) and biology. Whereas mothers appeared willing to take on this burden of genetic responsibility, in contrast, many described how ‘others’ within the family reject this possibility. Parents discuss how family members, particularly grandparents, react to the diagnosis of a genetic syndrome, highlighting the difficulties of transgenerational communication following diagnosis. “It’s not from my side” [Fam10] appears a significant feature in the parental story. The mother’s words below provide an example of how kinship can be reconstructed.

Researcher: Did your family know that you were going to go for testing?

Mother: Yes, actually that was quite, that was awful that was because I explained to them that if I had it, it would more than likely be run through the family, that made them angry. There is nothing wrong with us, you didn’t get it from us blah blah blah because [partner] made a comment about it one time and my dad went and bit his head off, there is nothing wrong with us, she didn’t get it from our side of the family and all I said was that you are not guaranteed that are you.

[Fam19 Interview with the mother of Rhian, diagnosed at 3 years old]

Whereas research often focuses on the dissemination of ‘risk’ information (Gaff et al., 2007) here it is the reaction of family members that draws attention. This mother highlighted how her father responded to the news, stating ‘there is nothing wrong with us’ and ‘she didn’t get it from our side of the family’. This extract therefore reveals that a diagnosis of a genetic disease has the potential for creating division within the family. The use of ‘us’ and ‘our’ are significant. ‘Sides’ are identified which map onto individual blood lines, and distinguished from the ‘other’. Blame and responsibility are negotiated by making visible one’s own

genetic heritage. Within this context, the head of the household appears to take up the role of ‘bloodkeeper’, protecting personal family heritage from the stigma of tainted blood. Genetic tests are therefore simultaneously individualising and collectivising as the information revealed is constructed as relating to *only* one side of the family.

Many of the extracts used in this article highlight how parents can represent themselves as responsible by being ‘open’ to the possibility of being the source of their child’s illness. However, parent accounts reveal that willingness to accept a role in genetic transmission is conditional. Blame is resolutely rejected when it is thrust upon parents from others in the family. The threat of blame appears particularly prominent when accounting for the behaviour of ‘in-laws’. This was the case for one mother who identified how communication within the family about the genetic diagnosis was accompanied by suspicion.

Researcher: Do you remember telling your family that it might be a family genetic syndrome?

Mother: Yes. And that’s when sort it sort of came out well, “it’s not from my side”.

Researcher: Who says that?

Mother: Husband’s side, yeah.

Researcher: How did you cope with that?

Mother: Er, it was quite hard really because, you know, because there was no proof either way to be sort of accu- it was almost as if we were accused. You know, it’s my - from my side and it’s my fault. You know, so that was very hard. So it was obviously, like I said it was a relief when we knew that it wasn’t from either side.

Researcher: How did you feel about being tested at the time?

Mother: I think it sort of was a relief really because everybody's - there was a lot of - from my husband's side of the family, well, it's not our side, it hasn't come from our side you know, and I was, you know, I was quite open minded. I didn't know where it had come from and why she had it.

[Fam10 Interview with the mother of April, diagnosed at one month old]

The rhetoric of blame and responsibility are used carefully in these parental narratives. When discussing a child's genetic illness it is acceptable to be 'open minded' for oneself while suggesting that other family members are seeking to lay blame. The 'other' that is dismissed in this way is often distanced from the parent, such as in-laws, or as one mother highlighted, an ex-husband "he would have fobbed it off, that if it was genetic it certainly wasn't from him" [Fam30]. Dismissal of the views of others as ignorant has been identified as one way in which potential stigma can be managed (Etchegary, 2010). The stories parents told about communication patterns suggest how a genetic diagnosis can be negotiated as a stigmatised identity. Stigma is a process whereby particular characteristics of a person or group are distinguished and 'discredited'. Examples of stigmatised illness include epilepsy or HIV, which might involve practices of concealment and restricted disclosure (Schneider and Conrad, 1980; Green, 2003). Whereas stigma is often associated with the reactions of 'outsiders' (Green, 2003), here 22q11 deletion syndrome appears to be stigmatised from within the family. Stigma therefore becomes part of, and is managed within, these 'webs' of kinship.

Managing stigma through protection and rejection is a significant determinant of practice and the enactment of family relationships. Genetic gatekeeping is about containment, and one example of this is in the re-writing of family history. The reproduction of the family narrative

is a reflexive process. The following account highlights how stories were told about family members taking active steps to rewrite their family history to ensure they were not implicated in the search for genetic cause.

Researcher: Were both your families involved in the discussions about the diagnosis?

Mother: Yes, they were...

Father: They were all terrified weren't they that they were somehow going to be responsible. They were all trying to distance themselves, in a way, in that they were trying to ensure that they...

Mother: Interesting over the years as well how much the various members of the family, some more than others, but both sides, but some a bit more than others, actually altered their medical histories a little.

Father: Yes. So the medical history we gave Dr Coombs is not the medical history that we now have. As it happens, there is no - there is nothing. But they were all so - they were all so clearly paranoid that somehow this defect was due to their side of the family. It was quite pathetic really.

Mother: And over the years little things have come out about oh, your uncle died of this, this and this.

Father: Congenital heart disease and somebody else had -

Mother: Really? Oh, you didn't tell us that. Ah, when was that then? Things like that. All sorts of stuff like that. Oh, surely we told you? No.

[Fam22 Interview with the mother and father of Harry, diagnosed at two and a half years old]

Within the clinical consultation the documentation of family history is a commonly used device for tracing illness through the family and identifying those at genetic risk (Featherstone *et al.*, 2006). The family tree is a vital tool that transforms family relationships into genetic relationships (Nukaga & Cambrosio, 1997; Author XXXX) and in the context of a genetic syndrome, its documentation represents valuable knowledge for current and future generations. Yet the extract above demonstrates flexibility in the family narrative. The respondents' attempts to construct a functional account were thwarted by those eager to disguise their personal histories in order to protect themselves and their heritage.

In contrast to the potential for fatalism following a genetic diagnosis (Whitmarsh *et al.*, 2007), the re-writing of family demonstrates engagement. Family history becomes malleable, one version of a story which is told or re-told to play a particular role. It is constructed through individual or sometimes collectively sanctioned judgements about what symptoms, and what histories might be relevant or might incriminate. The rejection of blame and the transformation of history from potentially tainted to clean once again emphasises the importance of examining personal and collective responses to genetic disease.

## **Conclusion**

Diagnosis, genetic risk and carrier status are highly significant concepts within biomedical practice. Advances in genetic technology have redefined disease classifications, resulting in a separation of the diagnostic process from the experience of symptoms, thereby blurring the boundaries between health and illness (Hedgecoe, 2003; Kerr, 2004; Miller *et al.*, 2005; Whitmarsh *et al.*, 2007). This is a salient issue in the context of biomedical advances, with the expansion of genetic testing and greater accessibility to one's own personal genome. The

development of new sequencing technologies for example has the potential to radically alter the diagnostic landscape for patients and families (Arribas-Ayllon et al., 2011). The resulting complexity of genetic knowledge has long been recognised as a critical barrier to communication of genetic information. Understanding how parents and family members respond to a genetic diagnosis, and how this is transformed by a de novo event, is therefore important.

The evolution of 22q11 deletion syndrome offers a timely and compelling opportunity to examine the contexts and consequences of genetic diagnosis. 22q11 deletion syndrome has attracted extensive scientific interest over the last twenty years, particularly in relation to the development of genetic technologies (Shprintzen & Golding-Kushner, 2008, Navon & Shwed, 2012). However, this article did not aim to focus on new and exciting promises of genetic technology. Instead, it reminded the reader of the importance of understanding the grounded experience of living with a genetic syndrome and examining the contextual frameworks through which genetic knowledge is translated. For the parents involved in this study, the process of negotiating a de novo genetic diagnosis involved making sense of the meanings of ‘genetic’, contextualising new knowledge within established frameworks of understandings and managing the response of others. These are the practical realities that demonstrate ‘genetic responsibility’.

The concept and meanings of genetic cause, particularly of family associations are powerful tools for making sense of illness and negotiating blame (Nelkin & Lindee, 1995; Rosenthal et al., 2001; Hallowell et al., 2006; Arribas-Ayllon et al. 2008). This article has demonstrated that this remains the case, even when a genetic syndrome is caused by a de novo deletion. A diagnosis of a genetic syndrome, irrespective of the pattern of hereditary, has personal and

collective implications. This contrasts with current approach that assumes that a de novo event is either irrelevant for family members (Leonard and Newton, 2010) or as McLaughlin & Clavering (2011) highlighted, is negotiated as ‘difference’ rather than ‘sameness’.

‘Family’ is a significant theme throughout this article. The relationship between a genetic diagnosis and family can be experienced as a burden, involving the negotiation of blame, rejection and protection. This article suggests flexibility in the role of ‘genetic house-keeping’. Mothers can accept responsibility and protect family members, yet significantly, responsibility can be rejected when blame is thrust from ‘others’ in the family. To know or not to know has emerged as a key question within the genetic age (Hallowell, 1999; Weiner, 2010) and parents are able to protect themselves from accusations because they did not, and could not have known about the risk. The de novo deletion is therefore used as a tool to refute genetic responsibility. However, a genetic diagnosis can also be valued because of the potential to reduce stigma (Phelan, 2002) and answer the question ‘why me?’ (Finkler, 2000). This article demonstrates that it is not just diagnosis that is important but also its mode of transmission. The genetic nature of the syndrome can be embraced because transmission from parent to child son offers hope that the child would enjoy the same quality of life. Here, hope is not invested in technological advances but in familiarity.

The responses of some parents in their attempts to understand cause and effect might appear proof of the limitations of ‘lay’ knowledge (Prior, 2003). Although patients might demonstrate experiential knowledge, Prior warned that “experience on its own is rarely sufficient to understand the technical complexities of disease causation, its consequences and management” (2003a p53). However, in drawing attention to these occasions, this article provides a valuable insight for understanding how genetic ideas are manipulated and

extended. This article confirms the multiple meanings of biomedical information, but most importantly, the significance of personal and familiar frameworks of knowledge for its translation (Parsons & Atkinson, 1992; Michie & Marteau, 1996).

I acknowledge that this study has several limitations. This study does not necessarily produce results that can be generalizable to a wider population. 22q11 deletion syndrome is a rare disease and the parents who contributed to this study may not share the same experiences as parents of children with different diseases. Likewise, as 22q11 deletion syndrome is associated with considerable variation, parents of children with the syndrome may themselves not share similar experiences. However, in light of the limited knowledge of how families negotiate a de novo genetic event, this article contributes rich detail about how and when family connections can become a burden or a resource. By contributing to this study, parents displayed a willingness to engage in discussion about their child's health. Those who are unwilling to talk about their experiences represent a hard to reach group and are not represented in this study. Therefore I would suggest researchers continue to explore diverse perspectives, including the ordinary and extraordinary, to enrich our understanding of the multiple implications of genetic knowledge.

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