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Voicing the lifeworld: Parental accounts of responsibility in genetic consultations for polycystic kidney disease

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ABSTRACT

When parents, who are carriers of or are affected by a genetic disorder, make decisions about the health risks faced by their children, there may be multiple factors to consider. These may include the medical benefits, the parents' own experiences of learning about their genetic status, and the future autonomy of the child. Health professionals face the challenge of explaining the possible burdens as well as benefits of testing children, while promoting open communication within families about the risk of an inherited condition. While genetic consultations do not in themselves constitute decision making, parents nevertheless account for their actions and decisions in an attempt to display parental responsibility.

In this paper we explore the accounting practices of parents in genetic consultations, focusing on how they articulate their responsibility with regard to testing their at-risk children for autosomal dominant polycystic kidney disease (PKD) and the communication issues surrounding the testing process and the disclosing of test results. Based on eight audio-recorded and transcribed genetic consultations from the UK, and drawing upon rhetorical discourse analysis, our findings suggest that (i) parents tend to foreground their practical 'lifeworld' considerations to justify their decisional actions; and (ii) there is considerable variation in the ways in which parents respond to information and advice offered by the professionals. The affected parent often presents their lifeworld concerns as outweighing, at least for the present moment, the longer term health benefits that might accrue to their children.

Introduction

Communicative tensions between 'the voice of medicine' and 'the voice of the lifeworld' are a common feature of health care consultations (Mishler, 1984, Silverman, 1987). Such tensions become even more pronounced when a child is involved, with diminished or no participatory status; parents, as carers, then have to mediate the consultation by orienting to the best interests of the child. Previous interactional studies of paediatric consultations (e.g. Aronsson & Rundström, 1988; Stivers, 2001; Strong, 1979; Tannen & Wallat, 1983, Tates & Meeuwesen, 2000) have drawn attention to how the child's voice may become backgrounded as parents and professionals routinely appropriate the role of spokesperson.

The existence of a hereditary genetic disorder presents difficulties for affected individuals in their role as parents, within and outside the clinic. If the condition is not evident at birth but typically presents in adult life, there are decisions to be made as to whether, when and how they should inform their children about the inherited nature of the condition and the fact that they are at risk of it. In the case of polycystic kidney disease (PKD), those who have inherited the condition often remain healthy well into middle life or beyond. In such circumstances, there may be two, often rather distinct, decisions to be made: (i) whether the healthy child should be tested as a young child; and (ii) how to discuss the condition with the child when older – as an adolescent or young adult – so that the 'child' can learn about the condition and their risk of it and, if not tested already, make their own decision about testing. Parents make these decisions either through an explicit process of weighing up the different considerations (whether to test or not, or to pass on information or not) or by default, i.e., by not making a decision and so neither testing their child nor informing them of their risk. The latter course was noted as problematic some thirty years ago in an interview-based study of one family from North America (Manjoney & McKegnay, 1978). The two questions noted above touch on ethical issues that remain highly contentious in the professional sphere. The first relates to debates

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about the genetic testing of children and the second relates to discussions about the transmission of genetic information across the generations within families. Given the contemporary importance of these issues, we wished to see if the original findings from 1978 remained relevant and applicable to other families today, especially in the context of genetic consultations.

In this paper we consider both questions in relation to the concept of parental responsibility for inheritance of PKD. We explore the reasoning set out in parental accounts of responsible decisional action. Where their decisions and reasoning might expose them to criticism as acting irresponsibly, we demonstrate that parents justify their actions or mitigate the charge of irresponsibility by an appeal to lifeworld considerations, including both practical and emotional factors.

As a theoretical basis, we adopt a role-relational perspective on responsibility (Emmett, 1966), which is a complement to the knowledge-based agency-and-intentionality dimension usually associated with responsible action (Sarangi, 2010). In what follows, we first provide a broad context for predictive genetic testing for children vis-à-vis parental responsibility and outline the medical context of PKD. We then introduce the data and methods before offering our findings and analysis. In the discussion and conclusion section we draw attention to the challenges for healthcare professionals in the face of variable accounting practices of parents. We also discuss the difficulty of drawing conclusions about how parents make decisions from the accounts they give in clinic and the relevance of our findings for professionals.

Predictive genetic testing of children vis-à-vis parental responsibility

With the progress and profile of the Human Genome Project in the 1990s, social scientists commented upon the psychosocial issues emerging as human genetics was increasingly applied to clinical practice. MacIntyre (1997) set out the contrast between the 'discourse of great promise' about genetics and the 'discourse of concern', in her discussion of ways in which genetics might lead to social discrimination against those at risk of disease; she raised the possibility of a new category of social identity for those who were still healthy but destined to become unwell. Richards (1993) summarised some of the early experiences of predictive testing for late-onset neurodegenerative disorders, especially Huntington's disease (HD), pointing out the surprise of practitioners at the modest numbers of those at risk who opted for predictive testing compared to those who had said they would before testing became feasible. There are difficulties with estimating the proportion of at risk adults who do seek testing but the low uptake is not in doubt (Tassicker et al., 2009). Wertz (1992) raised a number of important issues including the need for privacy of genetic test results because of possible discrimination in employment or insurance; the importance of this has been especially clear in countries where healthcare is funded largely through private insurance (Taylor, Treloar, Barlow-Stewart, Stranger, & Otlowski, 2008). This is in addition to the impact of the genetic testing process and of the test result on variables such as anxiety, depression, distress on the one hand (Dudok DeWit, Duivenvoorden et al., 1998; Dudok DeWit, Tibben et al., 1998) and the more complex 'survivor guilt' reactions, leading to alterations in familial relationships (Brouwer-DudokdeWit et al., 2002; Sobel & Cowan, 2000), on the other. With regard to disclosure, the dilemma extends to the moral obligations experienced by family members for each other and the fine ethical balancing between the decision to generate genetic information for and about oneself and the responsibility to disclose it for the sake of others (Hallowell, 1999; Hallowell et al., 2003; Taylor 2004).

Additional issues arise when parents request the predictive genetic testing of their young children because the latter are unable to give consent on their own behalf. When a child is ill with a condition that could be genetic in origin, then diagnostic investigations might demonstrate that the underlying cause of the condition is genetic; such testing is diagnostic rather than predictive and is a clinical necessity. Equally, when an at-risk child stands to benefit from testing despite being asymptomatic, as when those with the relevant disease-associated mutation can access medically useful interventions, then again the test is clearly warranted. There may also be good grounds for testing if the condition often causes signs or symptoms in childhood, so that the family is not left in prolonged and distressing uncertainty about possible early signs of the condition. When these circumstances do not apply, however, a consensus has developed among genetic health professionals and family support groups in many western countries that the predictive genetic testing of children should generally not be performed because such testing without clear medical benefit at the time will prevent the child from making his or her own decision about testing when older; in addition, s/he will not have the same privacy as an adult would expect and there is the possibility that the behaviour of parents or others towards the child might unduly bias their upbringing, especially if there are several siblings and they do not all have the same result (Borry, Goffin, Nys, & Dierickx, 2008; Clinical Genetics Society Working Party, 1994; Dalby, 1995).

As identified by Manjoney and McKegnay (1978) in relation to PKD, the crucial difficulty experienced by parents in relation to decisions about testing and the disclosure of test results or genetic risk is in the transmission of information to their children. This difficulty arises in other disease contexts too, such as Huntington's Disease (HD) (Skirton, 1998), balanced chromosomal rearrangements (Barnes, 1998, Jolly, Parsons, & Clarke, 1998) and cystic fibrosis (Parsons, Clarke, & Bradley, 2003). Previous studies indicate that parents sometimes fail to pass on relevant information even to their adult children (Jarvinen et al., 1999). In addition to the children's age, pre-existing family patterns of communication and relationship also influence parental disclosure of their BRCA1/2 genetic test results (Tercyak, Peshkin, DeMarco, Brogan, & Lerman, 2002). We share the general practitioner perspective that parents should, in general, pass on to their children, by the time they are adult, the information they may require to make important decisions about their future health care and reproduction. We also acknowledge that this obligation may be difficult to fulfill for a number of 'lifeworld' reasons. Parents, when discussing family communication in a research interview setting, usually speak from this practitioner perspective of the 'responsible parent', in which passing information to children is accepted as the appropriate course of action even if it may be difficult in practice and so deferred, because the person to be told is not perceived as ready to cope with the information (Arribas-Ayllon, Sarangi, & Clarke, 2008a and 2008b). We suggest that parents may orient to the researcher in an interview much as if s/he were a professional and display 'responsibility' in accounting for their decisions in a similar fashion. One would anticipate, therefore, that similar tensions about disclosure of information might surface in the clinic setting, with the professional hoping to promote open communication and the family members feeling accountable for any failure to do so.

The medical context: polycystic kidney disease

Polycystic kidney disease is a highly variable disorder inherited as a Mendelian autosomal dominant trait. It affects about 1 in 1000 people but often causes no symptoms until middle life when chronic renal failure may develop or an acute presentation with coronary artery disease or cerebrovascular accident may occur; the