Exploring genetic responsibility for the self, family and kin in the case of hereditary raised cholesterol

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**Article Info**

*Article history:*
Available online 13 May 2010

**Keywords:**
Heart disease
Genetic responsibility
Familial hypercholesterolaemia
Predictive testing
Genetic identity
UK

**Abstract**

This paper explores the notion of genetic responsibility, i.e. the responsibility to know and manage one’s own genome for oneself and the sake of others, focusing particularly on responsibilities to family and kin. It also considers wider ideas about the emergence of new forms of biological subjectivities with which the concept of genetic responsibility is associated. The paper draws on a UK-based study concerned with lay constructions of familial hypercholesterolaemia (FH), a treatable inherited form of high cholesterol, which involved qualitative interviews with 31 people with the condition recruited through a specialist outpatient clinic. The paper is an attempt to open out discussions about the significance of genetic responsibility and biological subjectivity. I argue that in this study, FH was not associated with a notable family narrative of illness or a strongly defined specific disease community, and no clear sense emerged of obligations to kin or others derived through genetic risks or genetic connections. While responsibilities concerned with the welfare of oneself and one’s existing offspring were enunciated, obligations to other potential or actual kin, e.g. to tell and encourage kin to manage their risks, were much less clearly defined. Drawing on these findings, I start to address questions about the pervasiveness of genetic responsibility and genetic identity and the contexts in which they might be significant.

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**Introduction**

In the wake of the Human Genome Project, it has been proposed that a new kind of biological subjectivity has emerged in which responsibility for the self extends to knowing and managing the implications of one’s own genome. The terms ‘genetic responsibility’ or ‘genetic prudence’ (Hallowell, 1999; Kenen, 1994; Novas & Rose, 2000) have been coined to encapsulate this idea. It has emerged in the context of expectations of an increasingly important place for genetic information and technologies in the diagnosis and treatment of many diseases.

The idea of genetic responsibility or prudence implies two areas of action. First, there is an obligation to become informed about one’s genetic constitution and to undertake risk management to monitor and try to modulate one’s own genetic risks. This aspect can be thought of as a continuation of more general discourses of personal responsibilities to avoid illness though identifying health risks and taking preventative actions (Petersen & Lupton, 1996). Second, genetic responsibilities are extended towards other people, particularly family members.

Two approaches for thinking about genetic responsibility are discernable in the literature. First, the social theorists Nikolas Rose and Carlos Novas (Novas & Rose, 2000; Rose, 2001, 2006, 2007; Rose & Novas, 2004) have suggested a wide set of obligations flowing from genetic knowledge, concerning decisions about the whole scope of life plans, such as forming relationships, having children, and career choices. Furthermore, they connect the notion of genetic prudence to a broader set of claims about the emergence of new forms of biological citizenship and activism. Thus, they argue that genetic information is collectivising in two ways:

‘Choices about marriage, procreation, financial planning, inheritance, career and much more are made in a web of entanglements involving actual and potential kin, employers, partners and children. And ‘at risk’ individuals are joining into groups and organisations, not merely demanding public provision and rights, but making their own claims on the deployment of biomedical technologies and the direction of biomedical research’ (Rose, 2001: p. 19).

Their discussions have foregrounded the activities of certain patients’ groups in engaging with science and shaping research. Indeed, they suggest that, in some cases, active membership of a patient organisation can be reconfigured as a duty (Rose & Novas, 2004). This version of genetic responsibility, therefore, ascribes the
genetically at risk person obligations towards potential and actual kin, as well as a desire, if not a duty, to participate actively in groups formed around a shared biological identity. In formulating this conception of genetic responsibility, Novas and Rose have drawn mainly on data from online support group discussions, in particular concerning Huntington’s Disease. Nevertheless, Rose (2006) explicitly extends the notion beyond ‘rare’ genetic conditions, arguing that it also applies to susceptibility for more common conditions.

The second approach to genetic responsibility is provided by the detailed empirical investigations of medical sociologists, ethicists and discourse analysts such as Arribas-Ayllon, Sarangi, and Clarke (2008), d’Agincourt-Canning (2001), Hallowell (1999) and Polzer, Mercer, and Goel (2002). This body of work, which draws on detailed ethnographic study, often involving interviews, has focussed on people’s narratives about their deliberations whether to undertake genetic testing and their actions relating to this. They have discussed, in particular, the responsibility to disseminate information to relatives and encourage others to get tested, as well as to know and manage one’s own risks for the sake of others. This approach can be supplemented by the growing body of more clinically-oriented studies concerned with the communication of genetic information within families (see Gaff et al., 2007; Peterson, 2005). Studies within this second approach have tended to recruit participants from those attending clinical genetics services. Much of this research has focussed around the cases of hereditary breast and ovarian cancer (HBOC) and Huntington’s Disease.

These different approaches help to alert us to the different types of sites where genetic responsibility may be articulated, for example, in online support groups, in the research interview, and in connection with particular conditions. Koch and Svendsen (2005) suggest a further site, when they observe that an ethos of responsibility for self, family and society is encoded in clinical genetic practices and policies. In other words, genetic responsibility may be embedded in certain institutional or political settings. Their work also helps to draw our attention to clinical discourse, and to the possibility that different forms of clinical management might align with or influence people’s accounts of living with genetic information.

Given the potential influence of different regimes of clinical management, it is worth reiterating that studies of genetic responsibility have tended to draw on attendees of clinical genetic services, and to focus on conditions for which genetic risk information is available in the absence of prophylactic therapies. Nevertheless, many conditions thought to have an appreciable genetic component may be managed wholly outside of clinical genetics and prophylactic therapies may be available. These areas have been relatively neglected in the sociological literature. It is possible that the rationale for seeking genetic information and the responses evoked by such information may differ in these circumstances.

Returning to Rose and Novas’s claims about the emergence of new forms of activism based around shared biological identities, one can argue that these ideas were developed through a focus on certain and perhaps particularly interesting patients’ organisations (O’Donovan, 2007). Furthermore, work in this vein has inevitably tended to focus on those who do participate rather than those who do not. Questions remain about how broadly such ideas apply.

It is with these observations in mind that this paper explores the notion of genetic responsibility in the context of inherited high cholesterol, focusing specifically on the case of familial hypercholesterolaemia (FH). This relatively common and treatable condition, which is associated with increased risk of heart disease, was selected specifically for its comparatively mundane character, its management outside of clinical genetics and the availability of widely prescribed prophylactic therapies. The study from which data are derived involved interviews with ‘ordinary’ people with FH who attend an outpatient clinic in the north of England, as opposed to studying only those who are active support group members.

The paper focuses on responsibilities to other people, particularly family, kin and potential kin. I have discussed understandings of FH and responsibilities for oneself in detail elsewhere (Weiner, 2009a). I will argue that the genetic aspects of FH are not prominent in people’s accounts of their experiences and actions, and conclude that responses to inherited conditions may partly depend on the characteristics of the disease in question, such as its treatability, prevalence, the kind of clinical management and technologies involved and the nature of lay models of causation with which it is associated. The research suggests attention both to the specific characteristics of diseases and to the different possible sites of study when theorising about genetic responsibility.

The following section provides some brief details about FH and its clinical management, before the paper moves on to the study design and findings.

**Familial hypercholesterolaemia: clinical context**

FH is characterised as a dominant single gene condition leading to increased risk of early and severe coronary heart disease (CHD), and is thought to affect 1 in 500 people in the UK. It has been estimated that, without treatment, at least 50 per cent of men and 30 per cent of women with FH would have heart disease or have died of heart disease by the age of 60 (Marks, Thorogood, Neil, Humphries, & Neil, 2006). The condition is treated mainly using the class of cholesterol lowering drugs known as statins, combined with dietary and other lifestyle modifications. Mortality rates for FH have fallen significantly since the introduction of statins in the 1990s.

In the UK, FH is currently diagnosed mainly on the basis of a set of clinical indicators including high blood cholesterol, family history and the possible presence of tendon xanthomas (cholesterol deposits or ‘lumps’ on tendons). Specialist care for people with FH is provided within the UK’s National Health Service through lipid clinics. These specialist outpatient clinics provide services for all people with lipid-related disorders, including, but not limited to, hereditary conditions. They are run by clinicians from a number of backgrounds, including lipidology, diabetology and clinical pathology.

It may be helpful to clarify the ways in which the management of FH is similar or different to ‘ordinary’ raised cholesterol in the UK. The clinical literature suggests that: (1) There is a large area of crossover in the cholesterol levels of people with and without FH, although some people with FH have exceptionally high cholesterol levels. (2) At similar cholesterol levels, people with FH are thought to be at greater risk of CHD than others, because they have had these high levels from birth. (3) Clinicians are urged to manage FH more intensively and treat it more aggressively than ‘ordinary’ high cholesterol, because of the higher risks of CHD. This means, for example, prescribing higher doses of statins, management via specialist clinicians rather than general practice, and more vigilant monitoring of cholesterol levels and other cardio-vascular risk factors.

**Methods**

This paper discusses data derived from a qualitative study concerned with lay and professional constructions of familial hypercholesterolaemia. It draws mainly on data from interviews with people with FH, who were recruited through a large lipid clinic in the north of England.
Recruitment and interview process

Following approval by the relevant NHS local research ethics committee, potential participants were selected randomly from those people aged 18 or over, whose diagnosis had been established for at least six months, who were due to have an outpatients appointment at the clinic between January and August 2004. People were invited into the study by letter, in advance of their scheduled appointments and about 60 percent of these agreed to participate. I undertook a total of 31 interviews. One was conducted at the interviewee’s home and all the remaining were undertaken at the hospital. I developed an interview topic guide based on the wider study aims and drawing particularly on the literature on genetic responsibility and lay constructions of genetic disease or susceptibility. This focussed on interviewees’ explanations of FH and CHD and their experiences and actions in relation to FH — see Table 1 (for further details, see Weiner, 2009a). The topic guide was used in a flexible way, and interviews covered some or all of the areas depending on the direction of discussion and on any time constraints indicated by the interviewee. Interviews lasted between 20 min and one and a half hours. I recorded all the interviews on a digital audio recorder and transcribed these in full.

Analysis

I undertook a thematic analysis of the interview data following the process outlined by Hammersley and Atkinson (1995), which involved an iterative process of identifying data that were relevant to existing themes in the literature, and identifying new themes through the identification of recurring talk on particular topics, recurrent use of a particular phrase, and attending to surprising or apparently inconsistent aspects. In this paper, I concentrate mainly on interviewees’ talk about their actions in relation to others and the sense in which obligations are derived through genetic knowledge or connection. In analysing the interviews, I have tried to avoid making assumptions about the veracity or otherwise of interviewees’ reported actions or thoughts. Rather, following Murphy (2000), the emphasis in the analysis is on what the interview data indicate about the normative frames associated with FH. In other words, what a person with FH might be expected to say in order to demonstrate that they behave normally, morally, and responsibly with respect to their condition. In the paper, interviewees are referred to throughout by an anonymised participant identification (ID) number.

Participants’ characteristics

The main demographic characteristics of the participants are shown in Table 2. All self-identified as white and the majority (28/31) as white British. Interviewees were all seasoned lipid clinic attendees whose diagnoses had been established for at least a year, if not substantially longer. According to their own

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<tr>
<th>Table 1</th>
<th>Interview topic guide — sample questions.</th>
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<tr>
<td>How did you come to be a patient at this clinic?</td>
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<td>What were your initial reactions to the diagnosis?</td>
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<td>Who did you tell about your diagnosis and what did you say?</td>
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<td>What ideas do you have about why you’ve got the heart problems/cholesterol problems/FH?</td>
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<td>If you had to explain your condition to someone else, what would you say?</td>
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<td>Are there any situations where you would prefer not to talk about it or avoid talking about it?</td>
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<td>What sort of impact has the condition had on your life?</td>
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<td>What would you say is the worst aspect of having this condition?</td>
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<th>Table 2</th>
<th>Sample characteristics.</th>
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<td>Gender</td>
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<tr>
<td>Male</td>
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<td>Female</td>
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<td>Age</td>
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<td>18–45</td>
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<td>46+</td>
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<td>Age range: 24–69 (mean 52)</td>
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<tr>
<td>Occupational background*</td>
<td>No</td>
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<td>managerial &amp; professional</td>
<td>15</td>
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<tr>
<td>Intermediate</td>
<td>7</td>
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<td>Routine &amp; manual</td>
<td>9</td>
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* Categories are drawn from the new National Statistics Socio-Economic Classifcation (Office for National Statistics, 2005). Intermediate occupations include clerical & technical occupations, and non-professional small employers & self-employed.

accounts, about half of the interviewees had already experienced some form of CHD, either heart attack or angina, and/or undergone some form of heart surgery.

Family narratives and a collective identity?

Previous studies such as Cox and McKellin (1999) and Richards (1996) suggest that, particularly for dominantly inherited genetic conditions, an awareness that the kinship may be ‘prone’ to the condition may become part of family culture; it becomes ‘this thing in our family’ (Cox & McKellin, 1999: p. 629). By contrast, people’s talk about their experiences of FH does not suggest a strong sense of collective identity based around the condition. For example, only a minority of the interviewees talked about a family narrative of a family history of CHD or raised cholesterol. In the current study, many interviewees either did not discuss family history or talked of piecing family health histories together after their own diagnosis was established. One woman, for example, said that she found out that she had raised cholesterol when she was thirty through having a ‘lump’ (tendon xanthoma) removed from her ankle, and was only later diagnosed with FH at the age of forty-four. She suggests that it was only at this point that she started to recognise the significance of her mother’s own health history:

ID11: my mother wasn’t one for talking a lot about things and she had big lumps on her hands which she wasn’t bothered about because they never hurt her. She never actually said she had high cholesterol, she just took these tablets that they gave her at the hospital. It was only when I started taking them that I realised that they were the same ones that she’d been on, but because by then she’d passed away.

In this research, many interviewees (60%) indicated that their own diagnosis of raised cholesterol had come about by chance or was unanticipated. Others (40%) decided to, or were advised to take a cholesterol test as a result of a relative’s CHD or high cholesterol. There is a distinction between this relatively large minority of people who suggested that family prompting or events had been the immediate precursor to them getting their own cholesterol tested, and the smaller subgroup of people who expressed a clear sense that heart disease or high cholesterol was a ‘thing in their family’. Prior to their own diagnosis, and even sometimes after this, interviewees did not necessarily have a strong sense of a family history of CHD. These observations are in keeping with earlier work on inherited cholesterol in the UK (Lambert & Rose, 1996), but are at odds with comparable research in the Netherlands (Horstman &
taking children, or persuading/encouraging adult offspring to attend for cholesterol testing in order to establish a diagnosis of FH, and encouraging appropriate behaviours and compliance with medication.

The diagnosis of children was, by and large, constructed as wholly obvious, straightforward and unproblematic; it was not framed as an action that would reveal something about one’s inner-self or identity, that might cause anxiety, or that might bring children prematurely into the medical sphere, but rather as a very practical act. Interviewees’ talk of their actions relating to their children focussed on being mindful of their children’s diets, and of encouraging or training their children to take their medication:

ID54: I wanted to make sure that all the family, the kids, if they needed the treatment they got it straight away and we make sure that they take their medication and I would think that they’re probably alright and live a normal life […] It’s because we’ve started them at a young age, it’s a habit […] I mean my children have got it and I don’t see how it will affect their lives at all, Dietary, I mean you’ve got to give some consideration to not taking the children to MacDonald’s everyday for their meals and you know we tend to eat lots of white meat […] It is noticeable in this extract that the interviewee connects his care of his children with the idea that they will probably have a normal life. This suggests that demonstrating responsibility for managing one’s children’s cholesterol, as well as constructing this as a largely pragmatic and unproblematic area, may be connected to warding off reproach for ‘passing on’ the condition in the first place.

Only one interviewee reported that she had declined testing of her child, because she wanted him to be able to be a child with ‘no labels’ attached:

ID100: He’s very active. He eats the diet that I eat and I just feel now he’s a child and I want him to be that with no label attached. And if he follows that regime for now, okay.

This woman’s account makes very clear that the fact her son had not been tested is not a matter of ignorance or fecklessness; this was a measured decision based on his psychological welfare and contingent upon his maintenance of appropriate behavioural regimes. In other words, this decision to decline testing can be seen as an act of parental responsibility.

Responsibilities to offspring continued into adulthood. A number of interviewees talked of trying to persuade or encourage their adult offspring to get tested, maintain healthy behavioural regimes and take their medication, or of their parents doing this for them. The welfare of one’s offspring is an unavoidably moral area (see Ribbens McCarthy, Edwards, & Gillies, 2000). These accounts suggest that people may take different approaches to discharging their duties to their offspring, but however this is achieved it is difficult to remain silent on this topic. Indeed, all but three interviewees discussed the diagnosis and care of offspring, of whom two did not have offspring. The prevalence of talk about actions in relation to one’s offspring may be an acknowledgement of the familial aspect of the condition. It is, however, hard to separate this from the responsibilities that any parent feels for the continued well-being of an offspring with any identified health-related issue.

Responsibility to siblings and wider kin

Previous studies such as d’Agincourt-Canning (2001), Hallowell (1999) and Polzer et al. (2002) suggest that obligations to communicate risk information and encourage risk management extend to siblings and wider kin. In the present study, talk on this
area assumed much less prominence than discussions about offspring. In keeping with previous studies, interviewees talked of informing those with whom they had social or geographical proximity. However, the ethical and psychological deliberations that seem to characterise accounts of communication in other studies (Featherstone, Atkinson, Bharadwaj, & Clarke, 2006; Gaff et al., 2007; Peterson, 2005) rarely featured in the current study.

Before providing further details, it is important to draw attention to certain methodological differences between the present study and those cited above. My analysis is not so much focussed on establishing the ‘facts’ or ‘realities’ of whether interviewees had or had not communicated with different kin and their reasons for this, but with the way this topic was (or was not) presented at the interview. It is concerned with whether interviewees need to demonstrate they have communicated with their kin in order to be seen by themselves and others (e.g. the interviewer) as proper and responsible people with FH (Murphy & Dingwall, 2003). This analytical stance, i.e. focusing on the moral work of interview talk, makes particular sense given that the interviewees were being asked to provide retrospective accounts of events and thoughts that were, in some cases, quite long past. I have characterised the types of narratives that emerged around this topic in three main ways:

**Claiming responsibility**

a small number of interviewees (4) readily credited themselves as having talked with kin or as being instrumental in others attending for testing. In the following example the interviewee can be seen to claim responsibility for, or attribute himself a central role in, the analytical stance, i.e. focusing on the moral work of interview talk, makes particular sense given that the interviewees were being asked to provide retrospective accounts of events and thoughts that were, in some cases, quite long past. I have characterised the types of narratives that emerged around this topic in three main ways:

**Not claiming responsibility**

a larger proportion of interviewees (perhaps a dozen) who mentioned talking with their kin about their condition did not obviously present themselves as being instrumental in their kin’s diagnosis or welfare, or claim agency in the same way as the interviewee quoted above. Their accounts of communicating with kin tended to involve less directive or first person language and sometimes emerged only through persistent questioning on my part. In the following account, for example, the interviewee’s talk suggests that he had a certain amount of discussion with his siblings about the raised cholesterol condition, but this was not framed as particularly urgent or significant, and these discussions were framed as more to do with accounting for his own CHD than concern for their welfare. It is through my persistent and rather single-minded questioning that he reiterates that he did speak to his siblings about the hereditary aspect of the condition:

KW: And so did you ever talk to them [brothers and sister] about it?
ID13: We did do, yeah, a long while ago but with marriage and children and things like that we only meet at weddings and funerals. So I don’t know how they’re going on. I think my sister attended, not here but I think she had a slight problem with cholesterol at one point.

**Not telling**

In a small number of cases (4), the interview data suggests that the interviewee had not talked with their kin about the condition. In one case the responsibility for telling kin was plainly refuted, and in part, this involved questioning the significance of the hereditary aspects, implying that other aetiologies may be involved:

KW: I was just wondering whether there’d been any kind of discussion with your brother?
ID88: no, we don’t particularly speak, you know, we don’t get on terribly well. […] But just as a note, my father died of a heart attack in 1965, so did his brother, okay. Now here’s the twist in this. His other brother is ninety four next month and has had angina for fifty years and never had an operation, and he’s still playing the accordion, the keyboard and a successful artist, now put that in your pipe and smoke it, init really? And his other brother is about eighty nine, so I dunno. There’s no answer is there?

In this case, it seems that the apparently sporadic, or unpredictable nature of heart disease was drawn on to lay off responsibility for talking with kin. This observation hints at a wider explanation that may contribute to the relative lack of prominence of this topic in these interviews. Earlier in the paper, I suggested that participants drew on many factors in accounting for their own or their relatives’ CHD. This finding is in keeping with other work on lay models of common diseases, even in cases where specific genetic susceptibilities have been identified, which report that genetic risks contribute just one aspect of multifactorial models (see Weiner, 2009a). In the case of CHD, while hereditary aspects may be acknowledged, individual experiences and behaviours tend to be prioritised. It is possible that in the case of FH, the availability of alternative explanations of heart disease may attenuate any responsibility people may feel to communicate with their families about the condition.

Overall then, only a small proportion of interviewees actively framed communicating with kin and encouraging them to manage their cholesterol as an obligation. Although many of them may have talked with their siblings or other family members about FH or raised cholesterol and know the status of these kin, family connections were not a prominent part of their talk. Occasionally, obligations based on such connections were actively rejected. The findings resonate with Horstman and Smand’s (2008) observation
that FH does not transform family ways of relating and that responses to the condition conform to existing family patterns.

Reproductive responsibility

Novas and Rose's notion of genetic prudence suggests that having knowledge of genetic risk information is associated with obligations regarding a number of areas of life planning. This section will focus on reproductive decision making, since, despite opportunity and prompting within the interview schedule, there was very little talk of other areas of life planning. One can argue that recourse to reproductive decision making, in particular, has become integral to the framing and management of genetic disease. In the UK, prenatal genetic testing and pre-implantation genetic diagnosis (PGD) are available for a range of Mendelian conditions including inherited susceptibility to certain cancers. By contrast, this theme was unimportant to the people with FH interviewed in this study.

Discussions about reproductive decisions were mostly prompted by me and were almost universally framed by interviewees as a non-topic, i.e. they had been contemplated, but quickly dismissed, or were outside the bounds of what is thinkable about FH. Interviewees, with only one exception, rejected the idea of reproductive decision making in connection with FH. This rejection hinged on the construction of FH as treatable, manageable and not serious enough to warrant such a course of action. The treatability and relative lack of impact of FH were underscored through comparisons with other, more serious conditions such as Huntington's Disease.

Furthermore, there was an explicit rejection of blame for transmitting FH between generations on the basis that (1) the condition is manageable and (2) that passing it on was an unavoidable result of chance:

KW: I’m wondering what ideas do you have about why you got FH?
ID67: It’s just the cards I was dealt. You know, that’s the way it is. It’s like, I’m about to have a child. The FH hasn’t affected that, you know, it’s a gamble. The baby may or may not have it and, I mean it’s not a debilitating condition. You know I'm perfectly healthy as I am now, and there’s treatment for it, so I don’t apportion any kind of blame or any guilt or anything like that. It’s just one of those things.

The lack of blame depends on a construction of FH as not only manageable and compatible with being ‘perfectly healthy’, but also as ‘a gamble’ and ‘just one of those things’. This construction of FH as unavoidable is striking given the focus on reproductive decision making in clinical genetic practices and policies. In other contexts, reproduction is clearly framed as a calculated and controlled decision, and as a responsibility.

One might argue that this rejection of reproductive decision making is quite understandable, given the treatable nature of the condition, and that conditions such as Huntington's Disease are indeed of a different order of seriousness. There is a danger, however, of being too essentialist about the specific characteristics of conditions. Petersen (2006) shows that people with classical genetic conditions, such as haemophilia, may make similar appeals to the manageability of their condition in their accounts of their reproductive decision making or make comparisons with other, ostensibly more serious conditions. He comments that these are, in fact, very familiar narratives concerning chronic illness more generally. Furthermore, like the present study, Hallowell et al. (2006) argue that, in the case of HBOC (a condition for which PGD is now licensed in the UK), men who are carriers of a BRCA breast cancer susceptibility gene may draw on a discourse of fate in order to present themselves as blameless with respect to the transmission of the gene. Such studies raise doubts about people's preparedness to guide their life decisions according to a particular biological subjectivity.

What possibly distinguishes the current study from these studies is not the narratives or actual actions reported by interviewees (e.g. appeals to fate, comparisons with other conditions, rejection of prenatal testing), but the way in which these are discussed. In contrast to studies of more classical genetic conditions (see also Downing, 2005), participants in the present study appeared to feel less obliged to say that they had deliberated about reproductive decisions, or perhaps, as discussed in the previous section, communicating with kin. This suggests that they may be subject to a different set of moral expectations, which may in part be related to professional and policy discourses associated with the conditions.

Discussion

The data I have presented suggest that genetic responsibility might be a fuzzier concept than it first appears and brings new perspectives to the notion. I have suggested that FH may not be associated with a notable family narrative of illness or specific disease identity or community. While all the participants were engaged in managing their condition, this cannot be thought of as prompted by a desire to know and manage their genetic constitution, since diagnosis often came about through happenstance and was established using clinical rather than genetic diagnostic techniques, and interviewees' accounts of FH were shaped by established ideas about CHD. Responsibilities regarding care of offspring, in terms of establishing diagnosis and training or encouraging children to manage their condition, were clearly enunciated. Talking with other kin and encouraging them to manage their risks was not a prominent part of these interviews, despite ample opportunity and prompting to discuss this area. Reproductive decision making was mainly seen as a non-topical, in other words as an idea which fell outside what is thinkable about FH. Overall, while these accounts construct a strong sense of obligation concerning the welfare of oneself and one's existing offspring, obligations to other kin, including future offspring, or a wider community of people with FH were much less clearly defined. The forms of collectivisation envisaged by Rose and Novas, in terms of life planning and involvement in lay health organisations, are virtually absent in these accounts of people with FH.

What does this analysis add to our understanding of genetic responsibility? It seems that the concept, particularly as formulated by Rose (2006), allows little space for the role of lay ideas about disease aetiology and the variable importance placed on the contribution of genes. Furthermore, it seems to rest on a model of genetic disease in which genetic risk information is provided in the absence of other biomarkers of bodily status or effective prophylactic treatments. It therefore does not anticipate the difficulties in the case of FH of distinguishing between genetic information and other health risk information and between genetic responsibilities and more general responsibilities to self and offspring. The present study suggests that where other aetiological explanations, other diagnostic technologies, and treatments are available, talk of life planning and responsibilities to kin may have less immediate resonance.

One might ask how specific these findings are to FH, and the degree to which more general conclusions can be drawn. It is possible that FH or conditions related to heart disease might be exceptional. Allsop, Jones, and Baggott's (2004) study of health consumer groups in the UK provides some evidence to support this idea, finding that there are comparatively fewer high profile groups
formed by patients or carers in this area. They comment (ibid: 744) that: ‘[i]t does not appear to arouse feeling of anger or resentment, or pose a threat to identity as the conditions mentioned previously [maternity services, mental health, cancer, arthritis]’ and suggest this might relate to ‘the high incidence of heart disease, the age and sex of those affected, the known links between lifestyle and the disease, and to the forms of clinical intervention available’. In short, there may attributes of particular conditions that make them more, or less, likely to become part of identity practices.

Drawing on the above study and the current analysis, one can suggest that heart disease may be distinguished particularly by (1) lay narratives about the condition being especially individualized and moralized; (2) the perceived ordinariness and ubiquity of the disease; and (3) the availability of both prophylactic and curative interventions. These may account, to some extent, for the lack of family narratives about heart disease and the absence of genetic responsibility observed in this study.

Nevertheless, on the basis of other recent studies concerning genetic susceptibility for adult onset conditions such as polycystic kidney disease (PKD) and Alzheimer’s Disease, it seems unlikely that people will adopt strongly defined genetic subjectivities for this category of conditions. In the case of PKD, for example, Cox and Starzomski (2003) argue that hereditary aspects are not at the forefront of people’s accounts of the condition, and the availability of preventative measures and non-genetic diagnostic technologies (ultrasound in this case) detracted from the potential utility and significance of pre-symptomatic genetic information. Lock (2008: p. 72) has argued that for those involved in research concerning genetic susceptibility for Alzheimer’s Disease: ‘Individuals do not apparently adopt genetically informed identities, nor believe their futures to be profoundly changed from what they had already envisioned’. In keeping with the current findings, these studies suggest a far greater concern with the here and now of prevention, treatment and support, than with future potentialities.

I am, however, reluctant to draw too firm a boundary between these conditions and the ‘classic’ Mendelian conditions that have tended to be the focus of study. I have highlighted a number of parallels in the narratives of people with FH and these classic conditions about reproductive responsibilities, suggesting that even for the classic conditions people may be unwilling to guide their conduct according to genetic information. I further suggested that what may distinguish FH from these conditions is whether or not interviewees feel obliged to show that they had deliberately, regardless of the decision reached, and proposed that that this may relate to the different forms of professional discourse associated with the conditions. This is a question about the degree to which lay narratives reflect the clinical framing of the conditions and the attendant responsibilities inscribed (Koch & Svendsen, 2005). In this context, study of the clinical practices associated with FH would be highly pertinent, although not covered by the current research.

Overall, the paper raises questions about people’s preparedness to assume genetic identities and forms of responsibilities. I have suggested that this may depend, to some extent, on the specific characteristics of the condition in question, including the types of diagnostic technologies employed, the availability of prophylactic therapies, the prevalence of the condition, and the nature of lay models of aetiology and types of clinical discourse and practice with which it is associated. However, studies such as Downing (2005), Hallowell et al. (2006) and Petersen (2006) indicate that even for the classic Mendelian conditions the idea of genetic responsibility may have been overplayed. In order to get a clearer picture of the significance of this notion, it may be necessary for future studies to consider a wider range of conditions, be alive to the possible influence of different cultures of professional practice, and consider both people who do and do not participate in disease-related communities.

References


