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Current Narratives

Volume 1
Issue 2 *Embracing Multiple Dimensions*

Article 4

1-1-2010

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Recommended Citation
Herbert, Dilinie; Cregan, Kate; Street, Alison; Bankier, Agnes; and Komesaroff, Paul, The lived experiences of families and individuals affected by haemophilia in relation to the availability of genetic testing services, *Current Narratives*, 2, 2010, 27-43.
Available at:http://ro.uow.edu.au/currentnarratives/vol1/iss2/4

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Abstract
Ill health may be related to a combination of genetic and environmental factors. Haemophilia, a rare congenital bleeding disorder, predominantly affects males and females may be identified as carriers. Genetic testing is available for individuals and family members who are interested to know their predisposition to the condition Thirty-nine members of a cohesive haemophilia community in Victoria, Australia, were interviewed about their attitudes towards genetic testing. The transcripts were analysed using thematic and narrative analysis techniques. The themes reflected the meanings people attached both to the disease itself and to the use of genetic testing to detect it. Narrative analysis was then employed to investigate these patterns of meaning further. We identified three typical narratives models within this haemophilia community: those of a male with haemophilia, of a female carrier and of a female non-obligate carrier (female without a familial predisposition to haemophilia). Close examination revealed a distinct pattern where aspects of the narratives tended to 'cluster' according to thematic categories. While people in the haemophilia community are broadly in favour of genetic testing and genetic counselling, males with haemophilia have concerns that arise in relation to biological data banks, female carriers are cautious about antenatal testing and support greater communication of risk within families, and female non-obligate carriers are specially concerned about the safety of obstetric practices. The pattern of responses we have identified indicates that, despite the proliferation of issues and themes across the narratives, the number of possible personal narratives in which they are embedded is in fact quite limited. In this sense narrative analysis offers a supplementary dimension to thematic analysis in the elucidation of qualitative data.

Keywords
Genetic testing, haemophilia, medical ethics

This journal article is available in Current Narratives: http://ro.uow.edu.au/currentnarratives/vol1/iss2/4

The lived experiences of families and individuals affected by haemophilia in relation to the availability of genetic testing services

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Abstract: *Ill health may be related to a combination of genetic and environmental factors. Haemophilia, a rare congenital bleeding disorder, predominantly affects males and females may be identified as carriers. Genetic testing is available for individuals and family members who are interested to know their predisposition to the condition*

Thirty-nine members of a cohesive haemophilia community in Victoria, Australia, were interviewed about their attitudes towards genetic testing. The transcripts were analysed using thematic and narrative analysis techniques. The themes reflected the meanings people attached both to the disease itself and to the use of genetic testing to detect it. Narrative analysis was then employed to investigate these patterns of meaning further.

We identified three typical narratives models within this haemophilia community: those of a male with haemophilia, of a female carrier and of a female non-obligate carrier (female without a familial predisposition to haemophilia). Close examination revealed a distinct pattern where aspects of the narratives tended to 'cluster' according to thematic categories. While people in the haemophilia community are broadly in favour of genetic testing and genetic counselling, males with haemophilia have concerns that arise in relation to biological data banks, female carriers are cautious about antenatal testing and support greater

communication of risk within families, and female non-obligate carriers are specially concerned about the safety of obstetric practices. The pattern of responses we have identified indicates that, despite the proliferation of issues and themes across the narratives, the number of possible personal narratives in which they are embedded is in fact quite limited. In this sense narrative analysis offers a supplementary dimension to thematic analysis in the elucidation of qualitative data.

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Introduction

> *'Now, as I try to write about Damon's childhood, to recreate a time where his small life was crowded with events, these are hopelessly confused in my own memory.'* — Bryce Courtenay

In *April Fools Day*, Bryce Courtenay shares with us his son's story of living with haemophilia. In a perfectly constructed narrative we hear about the moment a routine blood test revealed a life-long genetic disorder. We are allowed access to the private memories a father has involving his son and the experiences that transformed both their lives. From the joys of family reunions to the tragic circumstances surrounding his son's death, the book reveals an everyday account of illness and its effect within the family (Courtenay 1993). Bryce Courtenay is a well-respected author who has written numerous books. His stories about triumph over tragedy and love have inspired a generation of people to embrace fictional work as another realm of reality. The prose that Courtenay uses is unlike the stories we tell ourselves or share with others: our narratives construct the identity that is ours alone.

Haemophilia is a historically well-documented disorder. It was recorded in the Talmud and other ancient texts: for example, mothers were forbidden to circumcise their sons if there was a history of bleeding within the family (Rosner 1969). Haemophilia has also been referred to as the 'Royal Disease' because of its prevalence amongst the descendents of Queen Victoria, the most famous haemophilia carrier. This led to the condition presenting in the Russian, Prussian and Spanish royal houses through inter-marriage.

The scientific aspects of haemophilia have been well described. It is an X-linked bleeding disorder found to affect males, and females can sometimes be identified as carriers (Ragni and Schlesinger 2002). Unlike other chronic congenital disorders, haemophilia can be treated effectively, in this case with replacement clotting factor therapy. The consequence of prolonged bleeding, the hallmark of this disease, is restricted movement of the joints. Historically, men with haemophilia experienced a poor quality of life. Nowadays with safe, reliable treatments and occupational therapy they can expect to survive to an age comparable with that of the general population.

Haemophilia is a unique congenital condition which has been discussed extensively in the scientific literature but not to the same extent in the social sciences. Inheritance is usually confined within families, although it is not uncommon for individuals to present

with irregular bleeding without a familial history. This moderate level of penetrance shows that people can be readily identified as 'haemophilic', 'carrier' and 'non-obligate carrier' (i.e. a female without a familial predisposition to haemophilia). Regardless of demographic differences, there are obvious characteristics that are unique to each of these social identities which are removed from the genotypic aspects. In this study we demonstrate the way in which these social identities are constructed as narratives in relation to people's attitudes towards, and experiences of, genetic testing services.[1]

Chronic illness and related disabilities can have a profound impact on the ways that people are able to articulate meaning in their lives. Limited movement in some joints, caused by prolonged bleeding, can restrict some men from fulfilling an overtly physical life. There have been several definitions provided of chronic illnesses such as haemophilia, but essentially the term refers to a persistent state where 'symptoms are enduring and long term' (Lee and Poole 2005: 347). The experience of illness is not limited to the disease pathology but extends to the impact it has on people's lives and on those of others around them.

The research data used in this study are part of a larger project, which encompassed a needs analysis of community knowledge and attitudes towards genetic testing and genetic counselling.[2] In this article, we hope to provide an insight into the lives of members of the haemophilia community about the future utilisation of genetic testing. Of particular interest is the novel approach we have used to examine the complexities in the responses to genetic testing through the application of narrative discourse methods.

Narratives and storytelling

Storytelling is the foundation of all human-to-human interaction, regardless of culture of ethnic group. Other species show aspects of socialisation similar to those in human communities but speech, and our ability to create narratives using it, set us apart from other species in the animal kingdom. The stories that people share with each other are not often written down and rarely have special significance to anyone except the people who have shared in the experience. In fact, the earliest genealogies were spoken within families and passed on in oral traditions over generations. Historically, people have used stories for a variety of reasons including, in the spiritual context and in ancient writings, to document and express both extraordinary events and everyday events through personal journals. The literature about narrative expression or storytelling is extensive and related to different contexts in the social sciences and humanities. Essentially, narrative inquiry sets out to capture how people give meaning to and make sense of experiences.

Two key terms which have drawn much attention and debate in this field of research are *narrative* and *story*. For example, Paley and Eva (2005) first coined the term 'narrative

[1] The study of molecular biology was a significant phase in the development of genetic technologies. Genetic testing is the clinical application of gene discovery for individuals and families to learn about their predisposition to disease. It can be applied in antenatal and adult settings. Antenatal testing can be done in the embryonic or foetal stages; upon a positive result a decisions needs to be made about whether the pregnancy is continued or terminated.

[2] This project was conducted during 2006-2007 outcomes of which also appear in Herbert (2007). Other publications include Thomas et al (2007) which appeared in *Haemophilia*.

vigilance' as a recommendation not to confuse narrative with story. According to their research, a story represents the 'interweaving of plot and character, whose organisation is designed to elicit a certain emotional response' (2005: 83), whereas a narrative can be identified on a scale, depending on the presentation of particular events, being causally related to other events (Paley and Eva 2005). However, there are others who argue that narrative and story are synonymous and continue to use them interchangeably (Gilbert 2002). In this article, we use the term narrative to refer to the re-construction of events that have occurred in the past as they progress in the present and are projected into the future, as re-told from the perspectives of people living with a chronic illness — in this case, haemophilia.

Jonathan Culler (1981: 169) points out 'that the theory of narrative requires the distinction between what [we] shall call "story" — a sequence of actions or events, conceived as independent of their manifestations in discourse — and what [we] shall call "discourse" — the discursive presentation or narration of events'. Furthermore there needs to be differentiation between narratives and non-narratives, for if 'narrative is defined as a representation of a series of events, then the analyst must be non-discursive, non-textual given, something which exists prior to and independently of narrative presentations and which the narrative then reports' (Culler 1981: 171). The concept of time and the temporal sequence of events is an important aspect in identifying a narrative. Consistent with Culler's approach, we are interested in the discourses around haemophilia, and more specifically how they are used by individuals in recognisable patterns to construct senses of identity. However, the chronological and causal connections between these events are not always obvious, as Mieke Bal (1997) argues. She claims that an earlier event is not implicitly better or worse than one which follows, and that such distinctions need to be evaluated on their own merits (Bal 1997). In common with Bal, we do not make value judgments about haemophiliacs' understanding of their condition as it presents as a narrative of lived experience over time. We therefore understand narrative as a discursive construction that encompasses a very personal compilation of events and experiences that have been interpreted through the lens of the lived realities of people with haemophilia or their family members.

Narrative medicine

'Narrative medicine' is a relatively new term which has been used to describe the ways in which doctors and patients interact with each other. More precisely, Alcauskas and Charon (2008: 891) explain in their key article on the subject (which refers specifically to neurology) that narrative medicine

> [r]ather than a specialty is a framework for clinical practice based on developing and utilizing this skill set. It is a way of approaching the clinical encounter that focuses on appreciating and reflecting on the patient's experience and the patient–physician relationship in order to improve both by building trust, developing empathy, and fostering a sense of shared responsibility in a patient's health.

The importance of the study of narrative in medicine can be seen in the way that new forms of narrative medicine have emerged, including 'emergency medicine narratives', 'clinical narratives' and 'out of hospital narratives' (Hawkins 2004).

Narrative research has been used extensively in nursing research (Emden 1998a, 1998b; McAllister 2001; Pellico and Chinn 2007), medical education (Carson 2001; Bleakley 2005; Kumagai 2008) and in other health-related settings (Riley and Hawe 2005; Sanders et al 2007). The value of this research has been to show that empirical research based on statistical conclusions is not the only way to broach issues of human importance in the community. Narrative research, and in particular illness narratives, can inform the practices of medical staff and how they relate to their patients and also can contribute to the education of future doctors. In education, researchers' outcomes can be used to engage with students on an appropriate level to nurture a more productive learning environment. It is our aim, through our study of the narratives of haemophiliacs, to make a contribution to this endeavour.

Method

The gene sequences causing haemophilia were identified in the Factor VIII and Factor IX genes, which had been discovered in 1984 (Vehar et al 1984) and 1981 (Choo et al 1982) respectively. Genetic testing subsequently became available for families with haemophilia in many countries around the world. The technology of gene identification has also improved over the years. In Australia, genetic testing is readily offered to all families with haemophilia; for families in the State of Victoria, genetic testing is funded by the State Government.

We were invited to compose a needs analysis of the haemophilia community in preparation for government funding being made available to the community. Previously, genetic testing was paid for by the haemophilia treatment centres in Victoria. Today, families seeking genetic services are referred to a specialised genetic counselling service before being referred for genetic testing. In most cases individuals can be directly tested following their genetic counselling session.

The rationale for undertaking this study was to improve knowledge about how people in the haemophilia community understand genetic testing and about their attitudes towards it. In particular, we were concerned to find out what other complementary services should be provided to families and individuals seeking genetic testing and the role of genetic counsellors in the whole process. It is our intention that this information will be used to enhance genetic health services for the community.

Data collection

We invited participation through broad community engagement. Researchers were actively involved in speaking at conferences and community meetings to inform and invite people with haemophilia and their families to be involved in our project. We approached thirty-nine members of the haemophilia community, all of whom freely volunteered their time to participate in an interview. They included males with haemophilia, female carriers and female non-obligate carriers and other family members. Further information was also provided at the two main haemophilia treatment centres located in major metropolitan hospitals accommodating children and adult haemophilia patients in Melbourne.

Utilising well-established qualitative research methods, semi-structured interviews were conducted around issues raised in the literature in the area of genetic testing and haemophilia. The questions asked covered experiences of haemophilia and the understanding of the nature and significance of genetic counselling and genetic testing. The questions were developed to provoke participants' feelings about the prospect of widespread genetic testing services and related informational resources. Approval for the conduct of the study was obtained from the Human Research Ethics Committees at Monash University, the Alfred Hospital, and the Royal Children's Hospital in Melbourne. All interviews were digitally recorded and transcribed verbatim. Names and other identifiable details were omitted to preserve the anonymity of participants (Liamputtong and Ezzy 2005). We performed two levels of data-analysis: first, thematic analysis using the constant comparative method and then narrative analysis.[3]

Data analysis

Thematic analysis is commonly used in community health research (Boutain and Spigner 2008). Our use of thematic analysis revealed some very interesting categorical themes to emerge from the data, some of which were subsequently published in the international journal *Haemophilia* (Thomas et al 2007). Briefly, these themes related to: the experiences of having a condition of genetic origin; haemophilia itself, including its effects and treatments; issues arising from genetic counselling and genetic testing; and outcomes and implications of genetic testing.

Our decision to use narrative analysis in our further interpretation of the data was motivated by a desire to identify the ways in which the thematic categories arose in the contexts of individual narratives. In particular, we were interested to identify the way people who occupied conventionally recognised social roles in the community shared their stories. For the purpose of our research we have used the term 'narrative model' to describe the broad discursive constructions of three such key social roles in the haemophilia community: those of a male with haemophilia, of a female carrier and of a female non-obligate carrier. The narrative analysis was used to capture, in a collective sense, how each narrative model of these roles is constructed in response to the experience of haemophilia and genetic testing.

In the narrative analysis, in addition to the discourse analysis described above, we also used holistic content analysis. Discourse analysis traditionally perceives 'language as constitutive of experience as opposed to being reflective or presentational' (Willig 1999 cited in Stickley et al 2007). Holistic content analysis is an extension of mainstream qualitative research methods involving thematic analysis which we employed purposefully to deconstruct the narratives to gain deeper insights into the lived experiences depicted in the three narrative models of interest (Ollerenshaw and Creswell 2002). Using the whole interview data for each model, we uncovered ways in which meaning was constructed. We used the technique to identify functional links between the various thematic categories as they manifest in the three narrative models and thereby to uncover each of the model's underlying thematic content. Specifically,

[3] There are several notable limitations of narrative research, such as ensuring that population sample is sufficiently represented in some sense. Broadly qualitative research addresses this issue by ensuring that a level of saturation is reached pending analysis. Further follow-up would also constitute a level of validation.

the narrative analysis was applied in order to identify, in context, the occurrence of the themes identified in the thematic analysis and to explicate their inter-relationships.

Results

The systematic comparison of the main themes in relation to the narrative analysis is presented below. We show the results in two ways: a description of the thematic areas of interest identified (Thomas et al 2007) and a systematic comparison of the main themes entered into a table for further clarification (see Table 1 below).

The three narrative models that were identified coincide with the three key roles in the haemophilia community mentioned previously: those of a male with haemophilia, of a female carrier and of a female non-obligate carrier. Interviews for the purpose of this project were conducted with the total sample of 39 people. Two thirds fitted clearly within one or other of these models. The same analytical techniques were used to describe the narrative model identified with a family member without any direct genotypic affiliation with haemophilia but caring for a person with the condition; however, for the purposes of this paper we have chosen to confine ourselves to those associated with the three roles described.

The broad outlines of the experiences associated with the three groups are well-described. While haemophilia is usually inherited within families, it is not uncommon for males to present with severe bruising and bleeding and later be diagnosed with haemophilia without a familial predisposition. Generally, boys with haemophilia experience a normal quality of life, depending on the availability and course of treatment at the time. Other family members — in particular parents of haemophilic boys — are often actively involved with their treatments and encourage a sense of independence. As noted above, prolonged bleeding into joints often causes early onset arthritis which restricts mobility, compromising work and school commitments. The most serious complication experienced by the haemophilia community in the past was contaminated factor replacement therapy, leading to infection with HIV and Hepatitis C and was an enormous concern for the haemophilia community in the 1980s. This was a terrible tragedy of which strong memories persist in the haemophilia community today.

Women who are directly related to a male with haemophilia can be identified as carriers. A female carrier can be further differentiated on the basis of her relationship to a male with haemophilia: daughters are referred to as ‘obligate carriers’, siblings and other distant relatives are ‘potential carriers’ and mothers of newly diagnosed males are ‘non-obligate carriers’. For girls, growing up in a family with haemophilia can be a deep and difficult experience. They learn very quickly the limitations haemophilia can impose on their fathers and brothers and they also develop strategies to cope with their personal roles as sisters and carers. In later years these women often have to make decisions about child bearing and genetic testing. Similarly, non-obligate carriers must make decisions about future pregnancies.

Themes previously identified from the thematic analysis

The thematic analysis provided the foundation for the narrative analysis in this study. Conventionally, thematic analysis requires in-depth reading and re-reading of each of the interview transcripts. Discussions amongst the research team revealed the final

series of thematic headings. Each of the thematic headings is presented below with a corresponding quote to illustrate the theme in the context of the interview data. These thematic groupings are given as illustrations of how we came to identify the clusters of responses that led us to define the patterned narratives around each of the key roles.

Experiences relating to having a condition of genetic origin
There are both positive and negative aspects to the experience of haemophilia. The positive aspects are connected to identity and uniqueness, which is entailed in the narrative of a person with haemophilia. Sometimes a disease with a genetic predisposition can cause a level of stigma and guilt.

> Obviously it was traumatic for my parents because … it was out of left field for them. [Male with haemophilia]

> It was a bit of a relief knowing that it wasn't cancer. We were thinking that this is a life-long thing that we have to deal with. [Female carrier]

> [The haematologist] believed that Michael had haemophilia A and that it was severe haemophilia A so needless to say my daughter was born the next day at somewhat of a panic hoping that you know she wasn't a boy. [Female non-obligate carrier]

Experiences relating to haemophilia itself, including its effects and treatment
Treatment and quality of life are connected with each other. The chronic nature of haemophilia and associated bleeding can be relieved with appropriate and timely treatments. Synthetic replacement clotting factor therapy was much safer than the infusions given previously, which had caused a high incidence of HIV/AIDS in the community (Ragni and Schlesinger 2002).

> The treatment regimes … were very brand new, I guess in a lot of ways it was hit and misses even as far as the experts were concerned. [Male with haemophilia]

> I mean, the hepatitis and the AIDS crisis has been like a, you know, whole new thing. It's kind of you live a war with haemophilia … The HIV and the hepatitis kind of made the battle bigger than you know you retreat and keep on with the war haemophilia … If you don't learn all you can about [it] … you don't cope with it as well. [Female non-obligate carrier]

Issues relating to the process of genetic counselling and testing
In general, people in the haemophilia community are in favour of genetic testing and accept genetic counselling. The confusion arises when considering the purpose of these services. The benefit of having genetic testing is to identify potential carriers within families. Genetic testing can also be used in antenatal settings and for obstetric management as expressed by most women with haemophilia in their families. With greater reproductive choice came the complication of how people were to negotiate decisions about the selection of embryos and pregnancy termination.

> you can um make a lot of decisions based on that [genetic testing] and I think it is very important um but also that you get up to date information about haemophilia and treatment issues and things like that so its not based on old um family experience. [Male with haemophilia]
>
> Just for interest's sake to see where it had come from we wanted answers. [Female carrier]
>
> I don't have an issue with being a carrier myself. I don't feel guilty or anything [laugh]. I think it would be nice to know where it started. [And,] whether it had definitely started with me. [Female carrier]
>
> In a way I think it was just easy for us to try and conceive normally and if the baby has haemophilia — Oh well that is fine rather than have to make those horrible decisions. [Female carrier]

Outcomes and implications of genetic testing

The responses offered for this section were limited because there seemed to still be a level of confusion surrounding genetic testing. Contrary to scientific belief — that widespread genetic testing may potentially resolve future generations of haemophilic males leading to dismemberment of the haemophilia community — it was most likely that people expressed that genetic testing would lead to greater informed choice and strengthen relationships within families and the broader community.

> I hate to see someone terminate based on their uncle that had haemophilia and he lived until he was forty and he was in pain all the time … Nowadays you can carry on with normal day-to-day activities quite comfortably and have full working lives and things like that. [Male with haemophilia]
>
> the longer you have got to prepare yourself for a potential haemophiliac child I think can only be a good thing. [Male with haemophilia]
>
> Yeah, I had her [participant's daughter] tested. I was too curious [laughs]. Everyone was like just wait till she has children and I just thought no I can't wait. I just need to know. We were pretty sure that she wouldn't be a carrier because she doesn't fit … Sure enough she wasn't and that was lovely. [Female carrier]
>
> If my nephew turned out to have haemophilia I don't know that my sister would have dealt with it particularly well. Only because I think that the guilt is really the wrong word because there really is no guilt attached to it or shouldn't be. But there is, it's a subconscious thing that does attach and I think she would have found it very difficult to cope. [Female non-obligate carrier]

Narrative analysis: comparative presentation of themes

The narrative analysis is presented in the form of a table, as shown below (Table1). In our introduction, we explained how we examined the narrative content of associated with each of the three key social roles, the male with haemophilia, the female carrier

Table 1: Individual narrative styles of man with haemophilia, female carrier and female non-carrier in relation to temporal categories.

Temporal categories	Narrative one: Male with haemophilia	Narrative two: Female carrier	Narrative three: Female non-carrier
1. Experiences relating to having a condition of genetic origin	Identity, rebellion and discovery	Transformation, disorientation and responsibility	(No haemophilia-related experiences)
2. Early life experiences	Restricted to the confines of the family unit and regional area	Reformed by expectations and family circumstances (pre-symptomatic)	Limited direct experience but familiar with pathology through school
3. Familial relationships (parent/child)	Close connectedness with primary carer and unrestricted in daily activities	Resilient within the family unit and close connection with son	Close connection with son and minimal interaction within the broader family
4. Familial relationships (between siblings)	Challenged	Competing for attention	Depreciated within the family unit due to limited acknowledgement.
5. Personal development (education/occupation/friendships)	Social disconnectedness in school/work	Challenged by identity and repercussions of future family planning decisions	Minimally impacted
6. Interactions with health professionals	In partnership with doctors and allied health	Inquisitive to become more informed about haemophilia and its implications	Challenging authority for greater independence
7. Perspectives on genetic testing	Apprehensive, sceptical but in favour of greater informed choice	Great appreciation, supportive but not inclined to have it herself	Greatly supportive, in favour of widespread testing within the community
8. Perspectives of genetic counselling	Useful resource for educational purposes	Useful resource for education purposes	Compulsory resource for everyone with a history of haemophilia
9. Differentiation of risk (HIV/AIDS and obstetric management)	HIV infection was a difficult time emotionally and physically	Comforted by safer treatment products and feel obstetric management is the most important aspect of widespread genetic testing in the community	Safer treatments are reassuring and obstetric management is of the utmost importance

and the female non-obligate carrier. Here we have deconstructed the four thematic headings we have just described into nine categories. These correspond to the temporal sequence of events, what Culler (1981) called the 'story' and to which we refer as 'temporal categories'. These, in turn, are related to our discursively constructed narratives. Each column systematically summarises the experiences of the narrative model in relation to the temporal category. Similarly, each row shows the diverse nature and unique style of the three narrative models.

The male with haemophilia is connected on a deeper level to the experience of haemophilia because, in some respects, it is a key element of his identity. For him, growing up can be described as a time of discovery and rebellion. Many young boys with haemophilia find it difficult to accept the limitations that the condition can sometimes inflict on their daily activities, especially their social interactions. They therefore become confined to routines set for them by family members and carers within their 'home' boundaries. Sometimes these relationships are tested, for example, when these boys want to challenge these confines and pursue activities which might aggravate their haemophilia such as contact sports and labour-intensive vocations. Throughout life, a strong bond is formed with the medical profession for the purpose of treatments, therapies and other social services. This trust had been deeply compromised when clotting factor therapy became contaminated and for many older-generation men these struggles still resonate with them when they express their attitudes towards genetic testing and counselling. This may lead to scepticism about biological data banks and storage of genetic testing results. For many men with haemophilia genetic technologies are useful resources to have available in the community but only for educating people in the community and wider society about the condition.

Female carriers usually learn about haemophilia at a very young age. In most circumstances this education leads to a transformation in how they conduct themselves throughout their lives. They are profoundly affected by the experiences that they see played out in their families. It is rare for a carrier to show symptoms of bleeding, although it does occasionally occur. One of the most important issues female carriers have to consider is that of family planning. Their relationships with the medical profession are highly valued for clinical and social support. Genetic testing and genetic counselling are viewed as an appropriate resource for the purposes of family planning but are rarely used to assist decision-making about termination of pregnancy. Instead, it is more common for carriers to use genetic testing to better manage their pregnancies at the time of delivery.

Female non-obligate carriers usually have little or no experience of or knowledge about haemophilia. Information about haemophilia is usually obtained upon diagnosis of haemophilia in the family. At the time of diagnosis they can be intrigued by the condition as well as apprehensive about the complications and challenges it might impose on them, their sons and other family members. The family unit becomes closer and more intimately connected as family members become better informed. A strong connection is made between the family and the medical profession because it provides information about new developments in the treatment of haemophilia and about genetic technologies. Genetic counselling, in particular, is seen as a useful resource because of the information it can provide about family planning and further testing of other family members. There is often an eagerness to learn more about genetic predispositions, leading to a widely held view that such services should be compulsory for everyone in

the wider community. Having said this, decisions about child bearing are rarely based on genetic testing results although they still do inform decisions about obstetric management issues.

Males with haemophilia are connected on a deeper level to the experiences and the narrative construction of haemophilia because the latter is intimately bound up with their identities. Female carriers and non-obligate carriers have different relationships with haemophilia which are less intense in relation to the disease pathology and more closely bound up with the effects and impact on the lives of their families and especially their sons. In the familial relationships we recognise a close connectedness amongst family members, especially for males with haemophilia. This connectedness is not as obvious for female carriers, who often develop a high degree of resilience in relation to the challenges facing their families.

We also found that the experiences and meanings of certain aspects of genetic counselling and genetic testing are similar for women in both roles. Of particular interest are the benefits for greater obstetric management during pregnancy, no doubt a reflection of the fact that the risks associated with a haemophilic birth are profound for both the mother and the child.

Summary of results

Our results show how the interviews delineated the three narrative models that pattern around the three social roles. The models highlight the complexities of decision-making experienced by males with haemophilia, female carriers and female non-obligate carriers. They show how haemophilia is experienced differently by different family members and in different social settings and yet how there are common and recurring themes. In all cases, the experience of haemophilia has imposed significant stress on the family, but this has usually led to closer, more productive and satisfying relationships. All three narrative models describe the experiences of haemophilia as a source both of pain and of a deep sense of richness and growth, despite the different contents of each narrative.

The diverse and unique styles of the narrative models highlight how the lived experience of each social role is associated with a characteristic cluster of themes within each temporal category. This clustering effect demonstrates the multiplicity of dimensions of the expression and negotiation of decisions about genetic testing attached to relatively mundane, specific sequences of events.

Discussion

Years before Bryce Courtney published his experience of haemophilia, Robert Massie (1968) recounted his experience of having a son with the condition. This was the inspiration for his epic novel *Nicholas and Alexandra*, which was later produced as an Academy Award–winning feature-length film. The story was compiled using the diaries, letters and memoirs of contemporaries connected to the Tsar and the Empress of Russia. Massie shared with us the misfortunes of the Russian royal family, including in particular Tsarevich Alexis, who was born with haemophilia. The themes that arise in this novel are not too far removed from the experiences of families today (Massie 1968). For example, Empress Alexandra's reaction to her son's haemophilia is evocatively described: 'From that moment she lived in a sunless world reserved for the

mothers of haemophiliacs' (Massie 1968: 145). Arguably, the Empress's experience falls within our thematic categories of the early life experiences of haemophilia.

Although the social aspects of haemophilia remain largely undocumented (Ljung et al 1995; Petersen 2006), its scientific aspects are now much better known than they were at the turn of the twentieth century. Widespread genetic testing for haemophilia has the potential to produce significant benefits for the haemophilia community. It may be of particular value for couples seeking information during pregnancy: for women within families where there is the likely risk that they could also carry a relevant genetic mutation; for men with haemophilia to preserve their genotype for the benefit of future generations. To ensure genetic testing is not neglected or used inappropriately people in the haemophilia community need to be fully informed about the risks and benefits of using the service. Public health messages are a useful source of information in this context. However, studies have found that while people generally are responsive to such information, it rarely contributes to significant behavioural change (Sanders et al 2007). Further research is needed to show the role of public health messages in relation to the uptake of genetic testing services.

There is an increasing tendency for research that informs public health policy to draw on narrative methodologies. According to Riley and Hawe (2005: 227), the colloquial use of 'narrative' and 'story' in health promotion began in the 1990s to inform 'reflective practices and methods of program evaluation which can [give] more control to research participants' (Riley and Hawe 2005: 227). We have used these terms, drawing on theories from linguistics and semiotics, to give a further inflection to analyses that are drawn from public health research.

Although some social research into the experiences of people with haemophilia has been undertaken (Petersen 2006), none has employed the kind of narrative perspective we have used. Unlike other rare, chronic illnesses, the genetic nature of haemophilia has been shown to influence people's interest in and uptake of various medical and predictive services, but not necessarily to use them to prevent births. A study of birth records of males with haemophilia and prenatal testing in Sweden over a twenty-year period showed that the incidence of haemophilic births had remained unchanged during this period (Ljung et al 1995). The authors of this study argued that the nature and experience of haemophilia within families is as important in decision-making about health care and future generations as the simple availability of testing services.

Our research focused on the attitudes and experiences of members of the haemophilia community towards genetic testing. The narrative analysis presented in this paper provides insights into the nuances of how people experience haemophilia and make decisions about genetic testing. We believe that it thereby makes possible another level of understanding of these experiences that may guide further research and deepen community understanding of the lived experience of haemophilia.

Examples of such insights include accounts of the difficulties experienced by boys with haemophilia growing up. The condition severely restricted their activities, often resulting in prolonged absenteeism from school or work. The processes described how they forged a sense of normality in later life consistent with what sociologist Arthur Frank (1995) termed a 'restitution' narrative. In relation specifically to genetic testing our study highlighted the caution of males with haemophilia about genetic testing and

biological data banks as a result of concerns about possible implications about their own identities.

Similarly, the narratives of female siblings, daughters or mothers of males with haemophilia highlight how learning about carrier status for a woman can be a moving and transforming experience. This experience invariably led to further reflection about the implications of the condition and often to an experience of disconnectedness from other members of their families. In the case of female non-obligate carriers this disconnectedness often resolved as they become increasingly active in the care of their sons. For many mothers the diagnosis of haemophilia was linked to a sense of self-blame and guilt (Hern et al 2006), although it is possible that as women have become more empowered and assume independent responsibility for the care of their sons this aspect of their experience may become less marked. The women generally perceived genetic testing as a source of greater reproductive choice and safer obstetric practices. The risks associated with instrumental delivery at birth were a key issue for the women represented in both narrative models. Genetic counselling was generally viewed as a novel health service which could provide information to members of the haemophilia community rather than as an opportunity to reflect more deeply on their own identities and relationships.

Our interviews with the participants were focused especially on genetic testing rather than on other topics of potential interest such as the experience of puberty and adolescence and individual spiritual lives. It is likely that extended conversations around these issues would have further enriched the content of the narrative clusters and deepened the understanding about how various life choices are made. This is a potential topic for future research.

Conclusion

Haemophilia is a well characterised medical condition which is associated with an elaborate biomedical discourse that precisely defines the clinical features, the underlying physiological and genetic mechanisms and the available treatments. At the same time, as with other chronic illnesses, the experiences of people touched by haemophilia are complex, heterogeneous and diverse. Through the application of narrative techniques our study has suggested that these manifold experiences do not arise haphazardly or through mere aleatory combinations of disconnected circumstances. Rather, they are systematically generated within life stories of the individuals concerned. Furthermore, despite the broad multiplicity of themes according to which these experiences may be characterised the number of narrative models available is limited. Our study highlights the way in which narrative analysis, by demonstrating the clustering of thematic categories, can assist with the understanding of the structures of meaning and experience within a community.

Narrative analysis is an important research methodology that can provide insights into the experiences of individuals within particular communities. An understanding of how people negotiate decisions about their health is important for the development of public health policy and clinical practice. People with haemophilia invariably have to address a complex array of issues relating to, among other things, personal experience, relationships with family, professional carers and others, and the changing nature of medical care. Accordingly, we believe that a multidisciplinary approach to the development of haemophilia care which includes the study of narratives can contribute

to enhancing such care and ensuring that it is more responsive to the needs of community members.

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