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ARTICLE

Unravelling fears of genetic discrimination: an exploratory study of Dutch HCM families in an era of genetic non-discrimination acts

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Since the 1990s, many countries in Europe and the United States have enacted genetic non-discrimination legislation to prevent people from deferring genetic tests for fear that insurers or employers would discriminate against them based on that information. Although evidence for genetic discrimination exists, little is known about the origins and backgrounds of fears of discrimination and how it affects decisions for uptake of genetic testing. The aim of this article is to gain a better understanding of these fears and its possible impact on the uptake of testing by studying the case of hypertrophic cardiomyopathy (HCM). In a qualitative study, we followed six Dutch extended families involved in genetic testing for HCM for three-and-a-half years. Semi-structured interviews were conducted with 57 members of these families. Based on the narratives of the families, we suggest that fears of discrimination have to be situated in the broader social and life-course context of family and kin. We describe the processes in which families developed meaningful interpretations of genetic discrimination and how these interpretations affected family members' decisions to undergo genetic testing. Our findings show that fears of genetic discrimination do not so much stem from the opportunity of genetic testing but much more from earlier experiences of discrimination of diseased family members. These results help identify the possible limitations of genetic non-discrimination regulations and provide direction to clinicians supporting their clients as they confront issues of genetic testing and genetic discrimination.
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INTRODUCTION

With the advent of the Human Genome Project in the 1990s, one of the most contentious topics in public policy debates on genetic testing has been the use of genetic information by insurance companies and/or employers. Because insurance applicants are obliged to make full disclosure of relevant information to private insurance companies, people feared that genetic testing may render individuals uninsurable, leading to a 'genetic underclass'.[1] The term 'genetic discrimination' was coined to refer to the (negative) perceived differential treatment of individuals or their family members based on presumed or actual genetic differences rather than physical characteristics.[2] The threat of genetic discrimination has hindered medical research; according to Francis Collins, 'Unless Americans are convinced that the information will not be used against them, the era of personalized medicine will never come to pass.'[3] These fears also appear to disrupt health-care delivery. To keep genetic information out of their medical records, and out of the hands of insurers and/or employers, patients sometimes refuse genetic testing that could benefit their health. Some of those who chose to undergo testing sometimes pay out of pocket or use assumed names to keep genetic information private.[4]

To manage concerns about potential misuses of genetic discrimination, policymakers and lawmakers worldwide have taken measures to 'prevent' genetic discrimination.[5] In Europe, the Council of Europe's 'Oviedo Convention of Human Rights and Biomedicine'[6] has clearly set the tone by prohibiting any form of discrimination against a person on grounds of his or her genetic heritage and restricting the use of genetic tests to health purposes or scientific research. Since 1997, a substantial number of the European countries have enacted genetic non-discrimination regulations. In the United States, the Genetic Information Nondiscrimination Act was signed into law in 2008 to provide protection against genetic discrimination for employment and health insurance.[7,8] These regulations should help alleviate public fear of genetic discrimination, enabling the progress of genetic research and use of genetics in clinical and preventive care.

Despite these significant legislative efforts to protect individuals from the potential of genetic discrimination, its nature and extent have largely been undocumented. Research that validates the claim that genetic discrimination is occurring has been limited, both in scope and design. Reports of genetic discrimination have been criticized for being anecdotal,[9] and allegations of discrimination have usually been based on the presence of disease in contrast to genetic predisposition.[2,10–14] The most comprehensive study of genetic discrimination in asymptomatic individuals has been completed in Australia.[15–18] It documents numerous cases of genetic discrimination, the majority of which relate to the insurance industry and employment relations. In a recent Canadian study, Bombard *et al*[19,20]

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explored concerns and experiences of genetic discrimination among asymptomatic individuals in Huntington disease families based on family history and genetic test results, and reported that nearly 40% of the participants experienced genetic discrimination in the social sphere or in the domains of insurance or employment. Although limited evidence for genetic discrimination is gradually being collected, still little is known about the origins and backgrounds of fears of genetic discrimination and about how individuals react to (the risk of) genetic discrimination. Building further on Bombard's pioneering research, this article will extend the research from a monogenic condition, which exemplifies the almost-certain character of genetic predictions (eg, Huntington disease), to hypertrophic cardiomyopathy (HCM), a disease for which the relationship between genotype and phenotype is much more complex.

HCM is considered the most common genetic cardiovascular disease (affecting 1 in 500 persons worldwide). The disease is associated with sudden cardiac death at young age (especially athletes), but also seen as an important cause of heart failure at a later age.[21,22] Geneticists have shown that several hundreds of mutations in at least 27 genes, mainly coding for sarcomere proteins, have a role in the manifestation of the disease.[23] Genetic testing, however, still does not guarantee a conclusive result; in about 40% of the families with HCM who undergo testing procedures, no mutation will be found. HCM follows an autosomal dominant inheritance pattern, which means that children of a parent who carries the mutation run a 50% risk of being a carrier as well. The meaning of being 'at-risk' or even 'affected', however, is uncertain, because of the variable expressivity as well as the variable penetrance of the disease. Even within one family, the expression of the disease can vary strongly; some carriers of a mutation demonstrate no clinical symptoms, whereas others have an increased thickness of the myocardium or severe arrhythmia and suffer from serious heart failure or die after a cardiac arrest. Currently there is no cure for HCM, but following lifestyle advice (such as refraining from competitive sports), echo- and electrocardiographic monitoring or an implantable cardioverter defibrillator may help to prevent a cardiac event. Taking certain medications or septal reduction therapy are both associated with functional improvement and long-term survival.[24] Geneticists as well as cardiologists often see the possibilities of predictive genetic testing as a way to assess the risks of the disease long before clinically manifest dysfunctions appear and, with that, as a way to 'control' the disease in asymptomatic relatives by close cardiological follow-up. The lack of interventions to prevent the development of HCM and the uncertain meaning of being 'at-risk' and even of being 'affected', however, seem to be barriers for the uptake of a genetic test.[23,25] This article demonstrates that the fear of genetic discrimination may also importantly affect decisions about genetic testing in families with HCM.

In contrast to the Huntington disease research, which was based in Canada, a country where there are no specific laws addressing the use of genetic information by insurers or employers,[19,20] our study situates the concerns of genetic discrimination in the context of the Netherlands, with existing protective legislation. The Dutch Medical Examination Act (MEA)[26] restricts private insurers and employers in requesting a genetic test and using genetic test results from individuals who want to obtain a civil employment contract, a pension or a life or disability insurance. The act states that – for life insurance below a predefined ceiling of 160 000 Euro – no questions may be asked about untreatable hereditary disease or about the results of genetic tests for such diseases in the applicant and his/her relatives, except in case of an already manifest disease. As HCM is an untreatable hereditary disease according to the MEA definitions, HCM mutation carriers are protected by the Act in the case of non-manifest disease and when requesting insurance below the predefined ceiling. However, if HCM applicants apply for insurance beyond the predefined ceiling, they will have to make full disclosure of their HCM risk, potentially resulting in an increased insurance premium. This means that the MEA does not 100% protect HCM mutation carriers in insurance and there is currently public debate on how to interpret the Act's definitions.[12,27] The aim of this article is not to investigate whether or not the Dutch insurers and/or employers actually violate the Act and perform 'genetic discrimination' but to have a better understanding of the origins and backgrounds of fears of genetic discrimination and its possible impact on the uptake of HCM genetic testing.

METHODS

This article discusses data derived from a broader empirical longitudinal qualitative study concerned with the way families involved in genetic testing for HCM deal with a disease running in the family and with genetic testing in their everyday lives. In previous articles,[28,29] we have described the processes of data collection and data analysis in detail; here we will give a summary related to the data to be reported in this article.

After approval from the local medical ethical committee, the clinical geneticist and cardiologist who participated in the research team helped to bring us into contact with six extended families living with HCM. To allow for diversity in the families, the clinicians were asked to recruit families with and without a genetic test result, with few and many family members, with severe as well as mild manifestations of the disease, and families where children had been genetically tested. As the physicians worked as specialists in one of the two cardiogenetic centres in the Netherlands, it was easy for them to recruit families for our study. All who were asked by the professionals readily agreed to join the study.

In semi-structured interviews, participants were asked to explain how they learned the disease was running in their family, to describe the impact of living with the disease and with genetic testing, and to go into their decision on the non/uptake of genetic testing. Particularly, they were encouraged to talk about their everyday experiences with the familial disease. In the interviews, when talking about the non/uptake of a genetic test, fear of discrimination in insurance and employment was often articulated. This was striking, considering that regulation has been enacted in the Netherlands since 1998 to protect against the use of genetic information in insurance and employment, in case of non-manifest disease and below a predefined ceiling.

The article's analysis largely draws on interviews with four of the families involved in our study. The four families presented here were all white Dutch families, who differed in terms of size, place of living, social-economic status, profession, and disease and genetic status. To demonstrate the processes that shaped fears of genetic discrimination, this article presents four case studies, in which we describe in depth how these fears were articulated, experienced and interpreted by these families. Pseudonyms have been used to protect the identity of the participants.

RESULTS

The Goldfield family: postponing genetic testing

In the Goldfield family, taking up genetic testing for HCM became an issue after a mutation had been identified in 53-year-old Emily. Initially, many family members showed enthusiasm about the possibility of genetic testing. In the family, they recalled that many had suffered from heart complaints and some had died of a sudden cardiac arrest. During the first interviews, hopes were articulated of genetic testing as a way of breaking the Goldfield's family tradition of illness. However, after the clinical geneticist's suggestion about possible problems of insurance in case of a positive test result, the initial enthusiasm seemed to have been remarkably diminished.

When discussing their decision to have a genetic test done, many family members referred to the story of Ted, one of Emily's cousins. Ted, diagnosed with clinically manifest HCM, had to pay an extremely

high premium for life insurance and it was uncertain how long he could have his job as a truck driver. His story had given rise to the family idea that as Goldfields, in having a disease running in the family, they should always take caution with insurers and employers. It became clear that the clinical geneticist had triggered pre-existing fears of discrimination; it was in the light of Ted's previous experiences with 'that devil of insurance' that the clinical geneticist's information was perceived as a warning. As Emily explained, it had resulted in the idea of being better off without a genetic diagnosis:

> 'You might be restricted in all your actions and refused by insurance; so what the eye doesn't see, the heart doesn't grieve over. If you really don't know about being predisposed to increased thickness of the myocardium, you don't commit insurance fraud when saying you are healthy.'

The effect was that hardly any in the Goldfield family decided to take a HCM genetic test. Instead, most family members took action to 'safeguard' their future, for instance by buying a house before doing a genetic test. Genetic testing was *postponed* as a strategy to avoid 'genetic discrimination'.

The Green family: regretting genetic testing

In this family, Karen, a teenager who had experienced thickness of the myocardium since a young age, recently had a genetic test done. The cardiologist had recommended this as part of the diagnostic process, when her echo had showed a considerable increase in the thickness of the myocardium. After a mutation for HCM had been identified, Karen's two elder brothers, aged 18 and 20, were also offered genetic testing. At this stage, the clinical geneticist's suggestion of possible difficulties with insurance stimulated the parents to postpone testing their sons:

> 'It is not allowed to be silent about DNA; if the insurance company does a check and asks the biobank for information, they might get into problems. So it is better to wait until they have bought a house.'

In the Green parents' stories, genetic testing emerged as something other than 'just' another diagnostic tool; it was associated with themes like privacy and the prospect of an 'open future'. Both parents articulated a fear of 'big brother is watching you' and worried about the family's genetic information falling into the hands of third parties. These fears not only resulted in the postponement of testing their two sons but also in strong feelings of *regret* about the decision to have Karen tested.

> 'You hope you are making progress, but we don't get any further. Actually it gave us a fright; we had a lot of troubles taking out funeral insurance for her.'

For the parents, genetic testing had 'deepened the hallmark' of cardiovascular disease in Karen, reinforcing the feeling that she would be a subject to discrimination throughout her life.

The Anderson family: fearing a genetic test result

When we first met the Anderson parents and their two sons Dave and Christian, aged 20 and 21, the boys had already been waiting for their genetic test results for a year. At that time, both the boys had gone to the hospital with heart problems in the same week, and both had been diagnosed with increased thickness of the myocardium. As this had reminded the cardiologist of the possibility of a genetic disease, they had blood drawn for a genetic test.

In the first interviews, Christian explained that, since the HCM diagnosis, he had been worrying about his future, especially considering the idea that because of his 'heart abnormality', employers would discriminate against him.

> 'If I apply for a job, I will have to tell employers that I suffer from this disease and I think they'll prefer others for the job.'

For more than a year, he had been using alcohol and recreational drugs as a way of coping with the frustration of living with limited future prospects. In a second interview with the parents – 1 year after the initial interview – they expressed their relief that Christian had gradually succeeded in keeping away from alcohol and drugs and, against his expectations, had found a job. However, they now started to fear the possible impact of Christian's forthcoming genetic test result, as the identification of a genetic mutation might 'push him off balance again'. Although Christian had regained his stability, genetic testing was considered as a way of 'shaking up his identity' again; a positive genetic test result of HCM might open up old wounds of dealing with the consequences of being ill.

The Anderson parents also started to worry about the test results in the context of having children:

> 'It's not just better for Christian himself to have no result, but also for the children he might have in the future. As long as you don't know anything about a gene, you don't have to tell anybody.'

Ignorance of the genetic test results of their sons' disease was considered an attempt to deny its 'genetic character'. Although their heart complaints could be viewed as genetic, as long as this had not been confirmed by the test, they felt there was still a possibility of 'keeping it silent' and away from third parties.

The Redford family: coping with genetic test results

Nick Redford, in his thirties, had had a sudden cardiac arrest at the age of 15. Although at that time, a DNA test was not available, family history and heart examinations of his father and brother had made clear that his complaints had a genetic character. Nick remembered that the HCM diagnosis, as well as the emphasis on a strict regime of limited physical exertion, had given him difficulties in the years he was finishing school and finding a job. Also later, when buying a house, he had experienced problems related to his disease, such as extremely high insurance premiums, and after the implantation of an implantable cardioverter defibrillator, he had no longer been permitted to drive a car for work purposes.

These negative experiences, however, had never held Nick back from seeking the latest biomedical knowledge for the future of his young twins, a boy and a girl. On the contrary, they resulted in high hopes for possible treatment that would enable his children to escape the family's biological fate. When the twins were 3-years-old, DNA testing for HCM became available. The children were tested and it turned out that one of them, the boy, had the mutation, whereas the girl did not. Although some in the family – particularly Nick's mother – expressed concern, because of a fear that 'there was already a mark on this boy' and that it was just the way that 'when filling out insurance forms, you needed to be honest', Nick did not experience any feelings of fear or regret at all. He referred to his experiences with living with the disease by emphasizing that he had always found ways in dealing with the problems with which he had been confronted. For example, to avoid issues with employment discrimination, he had started his own company. When Nick had to take out life insurance as

part of a mortgage, he had kept the amount insured below the predefined sum to prevent having to do a medical examination. Furthermore, in terms of preventing possible discrimination problems with his car insurance, Nick had his own specific strategy:

> 'I always have golf equipment in my car. If I have an accident when driving the car for work purposes, I can say that I am going to play golf and travel for private purpose'.

Nick's own preceding experiences, and the strategies he had developed to avoid discrimination, prompted the belief that he would also be able to help his son in successfully coping with the HCM mutation and its possible problems of discrimination.

DISCUSSION AND CONCLUSION

Our findings indicate that although Dutch policymakers try to prevent public fear of genetic discrimination by installing genetic non-discrimination regulations in the Netherlands, in practice, the HCM families that participated in our study were still concerned about insurance and employment discrimination. This study analyzes the processes that shape fear of genetic discrimination and provides an insight into the influence of at-risk families' perceptions of fears of genetic discrimination for the uptake of genetic testing. The results support previous research on the concerns and experiences of genetic discrimination in the context of Huntington disease in Canada,[20] but this study is the first to provide a detailed description of the social mechanisms of fears of genetic discrimination. Moreover, it addresses both a disease like HCM for which the relationship between genotype and phenotype is quite complex compared with monogenetic disorders and a national context where genetic non-discrimination regulation exists (Dutch MEA). Although it has been argued that fears of genetic discrimination will be eased because of the more uncertain character of many monogenic as well as multifactorial diseases,[30] this study highlights that families living with such an 'uncertain' disease also expressed fears of genetic discrimination in the contexts of insurance and employment. Additionally, the study indicates that the installation of genetic non-discrimination regulations does not appear to alleviate these concerns of genetic discrimination.

Most importantly, our findings suggest that fears of genetic discrimination in at-risk family-research participants of HCM did not result from the (the possibility of) genetic testing as such, but were primarily nested in pre-existing experiences of discrimination within these families. These 'family histories', encompassing shared experiences of discrimination, living with disease in the family, 'being a member of an affected family' and so on, seemed to affect the way family members decided to undergo or refrain from genetic testing. As such, when a clinician offers genetic testing for HCM, it may trigger memories of living with a cardiac disease that wreaks havoc in their family and the consequences of which they are all already too familiar with from early childhood. These pre-existing concerns of discrimination seemed to have an important role in justifying the non-uptake of genetic testing for the HCM family members and did not only result in postponement (eg, the Goldfield family) but also in feelings of regret over having the genetic test done (eg, the Green family) or the wish of never knowing the genetic test results (eg, the Anderson family).

So, to understand the non/uptake of genetic testing, the previous experiences of the families in coming to terms with their disease, and its societal consequences, must be taken into account.[31] In particular, our findings highlight the broader context of the family and 'family history' of living with HCM and the work that families have to do in coping with genetic testing and genetic discrimination in light of previous family experiences of discrimination. This attention to the way families have managed to live their lives with disease and the strategies developed to cope with discrimination or exclusion may also explain why some families (eg, the Redford family) develop a positive account of genetic testing for HCM. This study provides further insight into the impact of genetic testing on the family and highlights a need to pay special attention to the familial contexts during pretest and post-test counselling.[28,29] These results also suggest the need for further research on genetic discrimination in this area.

The findings of this study also indicate the limits of a narrow concept of 'genetic discrimination'. Genetic non-discrimination regulations that have been enacted to protect people from 'genetic discrimination' in order to enable the uptake of genetic testing tend to focus on the novelty and specifics of 'genetic discrimination'. However, when genetic testing entered the lives of the HCM families, these families already had previous experiences with 'disease in the family' and with related problems of discrimination. Although previous experiences such as these are not taken into account in these genetic-specific regulations, they do seem to affect decision-making about HCM genetic testing. In addition, the narratives of the families in this study seem to resonate a fear that is not directly related to exclusion from insurance or employment but to larger concerns of privacy protection and disclosure of issues that are preferred to be kept as 'family secrets'. Within such a view, the offer of genetic testing for HCM can be considered as a 'hallmark' of preceding concerns of discrimination of these 'family secrets'.

Further, the families in our study seemed to be concerned about the risk that their children may be discriminated against in future; this was the case even when these children were still unborn. In this regard, our study is instructive in that both 'asymptomatic individuals' and 'symptomatic individuals' expressed concerns of genetic discrimination. Although the Dutch MEA protects HCM mutation carriers in case of non-manifest disease (the healthy 'at-risk') from 'genetic discrimination', these people expressed fears of discrimination as a reason for non-uptake of genetic testing or regret for having the test done. It is remarkable that also those family members with already manifest symptoms of HCM, for whom genetic testing is much more a diagnostic tool than a predictive one, expressed fear. Because, these people already faced high insurance premiums or exclusion in employment because of disease. Notwithstanding the MEA, family members of patients were not confident that information about their genetic make-up would be kept private. In contrast, they genuinely worried about the genetic information falling into the hands of third parties such as insurers or employers, and resultantly, running into further difficulties. Although legal definitions of genetic non-discrimination regulations focus on the boundary between symptomatic and asymptomatic individuals in offering protection against genetic discrimination, in the experiences and interpretations of individuals and families at-risk, these boundaries were not so clear-cut. Recent studies on the prevalence and extent of genetic discrimination have only described that people experience genetic discrimination based on family history.[19] Our findings may help to understand how the (initial) reactions to and concerns of genetic discrimination indicate concerns, not only from asymptomatic individuals but also from symptomatic individuals.

Furthermore, these fears of genetic discrimination in the context of symptomatic individuals seem to resonate with broader experiences of stigmatization in living as a family with cardiac disease. This refers to more diffuse verdicts of social unworthiness, prejudice and

stereotypes towards their family, and with these families' coping with this kind of 'family picture'.[32] This study provides further insight into the role of these more 'indirect' forms of discrimination and broader experiences of stigmatization. It also highlights the need to pay special attention to the types of experiences that limit the choices and options available to persons or families 'at-risk'.[19,32,33] These results suggest the need for further research into how experiences of indirect forms of discrimination may affect concerns relating to genetic discrimination.

Finally, although the interpretation of the Dutch MEA has been a topic of debate in the Netherlands, for example in the context of two recent Dutch reports of discrimination experiences,[12,13] our study suggests that the causes of (persistent) fears of genetic discrimination should not be only confined to the definitions of the law and its difficult wording. Our study highlights that fears of genetic discrimination are often expressions of preceding experiences with living with the societal consequences of disease in families, where familial experience ranged from asymptomatic to symptomatic individuals and was based on their mutual interactions within the family. In other words, (persistent) fears of genetic discrimination of the people may have less to do with 'misunderstandings' of the law or 'insufficient information communication' about the workings of the law but may be rather nested into previous experiences of being a member of a 'family at-risk'.

Limitations of the study

In the interpretation of our findings, several considerations should be taken into account. First, the family members who agreed to participate in our study may be a self-selected group and may demonstrate specific perceptions and experiences of genetic testing and genetic discrimination. However, we did not study an extreme population. In the light of the variety of the characteristics of participants, we assume that these families and family members can be seen as representative for how HCM families experience genetic testing, and perceive and cope with the fear of genetic discrimination.

The data was based on experiences from a Dutch sample whose concerns and experiences may not apply to other populations, particularly in the light of the specific Dutch genetic non-discrimination regulations. In genetic counselling, however, it is the perceptions and fears of the clients that are important, and thus these research findings also provide clinicians in other countries a framework to understand and contextualize the experiences and fears that clients share with them. Nevertheless, different national styles in dealing with risks, and different cultural values with respect to genetic testing,[34–36] might have an effect on the way individuals as well as families experience genetic technology and the risks of genetic discrimination.

Finally, because the sample consisted of families in the trajectory of genetic testing for HCM, the study findings may not necessarily apply to other genetic and non-genetic populations.

Implications of our results

Our findings have implications for clinical geneticists, as well as cardiologists, who work with HCM families. In counselling about genetic testing for HCM, it is instructive for these professionals to acknowledge that providing information on genetic non-discrimination legislation may not be sufficient in justifying the decision of the HCM patients to non/uptake genetic testing. This may not be sufficient as justifications for genetic testing and fears of genetic discrimination may be nested in broader 'family histories' and narratives of discrimination. This implies that counselling sessions should be focusing less on the information provision of genetic non-discrimination legislation and more on the role of family dynamics and individual strategies to cope with the social consequences of living with HCM as possible barriers for uptake of genetic testing. By acknowledging the way genetic discrimination 'connects' with the broader 'family histories' of HCM families may be an important factor in realizing the potentials of genetic testing in clinical practice.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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