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Short Report

Managing genetic discrimination: Strategies used by individuals found to have the Huntington disease mutation

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The introduction of predictive testing for Huntington disease (HD) over 20 years ago has led to the advent of a new group of individuals found to have the HD mutation that are currently asymptomatic, yet destined in all likelihood to become affected at some point in the future. Genetic discrimination, a social risk associated with predictive testing, is the differential treatment of individuals based on genotypic difference rather than physical characteristics. While evidence for genetic discrimination exists, little is known about how individuals found to have the HD mutation cope with the potential for or experiences of genetic discrimination. The purpose of this study was to explore how individuals found to have the HD mutation manage the risk and experience of genetic discrimination. Semi-structured individual interviews were conducted with 37 individuals who were found to have the HD mutation and analysed using grounded theory methods. The findings suggest four main strategies: ''keeping low'', minimizing, pre-empting and confronting genetic discrimination. Strategies varied depending on individuals' level of engagement with genetic discrimination and the nature of the experience (actual experience of genetic discrimination or concern for its potential). This exploratory framework may explain the variation in approaches and reactions to genetic discrimination among individuals living with an increased risk for HD and may offer insight for persons at risk for other late-onset genetic diseases to cope with genetic discrimination.

Y Bombard[a], E Penziner[b], J Decolongon[c], MLN Klimek[d], S Creighton[e], O Suchowersky[d], M Guttman[f], JS Paulsen[b], JL Bottorff[g] and MR Hayden[a]

[a]Centre for Molecular Medicine and Therapeutics, Child and Family Research Institute, Department of Medical Genetics, University of British Columbia, Vancouver, BC, Canada, [b]Departments of Psychiatry, Neurology, Psychology and Neurosciences, University of Iowa, Iowa City, Iowa, USA, and [c]Huntington Disease Medical Clinic, Department of Medical Genetics, UBC Hospital, University of British Columbia, Vancouver, British Columbia, [d]Departments of Medical Genetics and Clinical Neurosciences, University of Calgary, Calgary Alberta, [e]Provincial Medical Genetics Programme, Children's and Women's Health Centre of British Columbia, Vancouver, [f]Centre for Movement Disorders, Markham, Ontario, and [g]Faculty of Health and Social Development, University of British Columbia Okanagan Kelowna, British Columbia, Canada

Corresponding author: Dr Michael R. Hayden, Centre for Molecular Medicine and Therapeutics, Child and Family Research Institute, Department of Medical Genetics, University of British Columbia, Room 3024–950, West 28th Avenue, Vancouver, British Columbia, V5Z 4H4 Canada.
Tel.: 604 875 3535;
fax: 604 875 3819;
e-mail: mrh@cmmt.ubc.ca

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Huntington disease (HD) is an autosomal-dominant neurodegenerative disorder that usually presents in adult life with cognitive, psychiatric and motor disturbances. HD has a prevalence of approximately 5–10 per 100, 000 and is inexorably progressive ending in death approximately 15–20 years from onset (1, 2). New approaches to treatment are being explored; however, a cure or therapy does not currently exist to alter the course of the illness.

The discovery of an expanded CAG trinucleotide repeat as the underlying mutation that causes HD led to the availability of direct predictive testing (3). The introduction of predictive testing for HD has led to a new group of individuals found to have the HD mutation who are currently asymptomatic, yet destined in all likelihood to become affected at some point in the future (4). Individuals identified with the HD mutation have a CAG repeat length of over 36 and are considered to be at 'increased risk' for developing HD in their lifetime, should they live long enough (5). In fact, there is a significant inverse relationship between CAG repeat lengths and age of onset of HD with a larger CAG expansion associated with an earlier age of onset (4). Some individuals with repeat lengths between 36 and 39 years may never develop symptoms of HD in their lifetime, even if they live to an advanced age as CAG expansions between 36 and 39 years are in the affected range, but are not fully penetrant (4, 6). While CAG length is the major determinant of the age of onset, other genetic and environmental factors are also likely to contribute to the variance in age of onset of HD (7).

The advent of predictive testing for HD introduced many opportunities as well as potential challenges for individuals at risk for HD. One potential consequence of predictive testing is genetic discrimination. Genetic discrimination (GD) refers to the differential treatment of individuals or their family members based on presumed or actual genotypic difference rather than phenotype (8).

There have been four reports of genetic discrimination in the context of HD (9–11). Kenen and Schmidt (1978) warned of the dangers of stigmatization of individuals found to have the HD mutation (9). Although testing was not available at that time, they speculated that at-risk individuals live their lives in a "suspect state", neither fully stigmatized nor "considered fully normal as the label of defective individual lurks in the background" (9). Predictive testing, they presumed, would replace this uncertainty with social stigma and limit opportunities of these individuals as a result of their future illness. A survey of 27,790 individuals and children at risk for genetic disorders discovered 276 reports of GD among the 623 HD respondents (10). Of the 206 follow-up verification interviews conducted five cases concerned discrimination against individuals at risk for HD. These reports included life insurance rejection, coercion by a physician to undergo prenatal genetic testing and abort an affected foetus, two cases of adoption denials and a job refusal for the US Air Force (10).

An informal poll in Canada found frequent unreported instances of genetic discrimination because of a family history of HD or based on predictive testing (12). Furthermore, three individuals reported employment difficulties following disclosure of their genetic test results to their employers (12). The case of a teacher in Germany who was refused a job because of being at risk for HD (11) lends credence to the potential for such discrimination in the HD community worldwide.

Fears of genetic discrimination have precipitated altered behaviour around predictive testing for HD including the request for anonymous predictive testing. Several individuals have sought predictive testing under anonymity in fear of genetic discrimination for themselves and their families in Canada, the United States and Europe (13–16). The selective disclosure of genetic risk information for fear of discrimination has also been reported as another strategy to avoid potential loss of opportunities in the employment context (17). Finally, withholding information about seeking HD predictive testing from health-care providers has been previously shown as another method of limiting insurance or employment discrimination (18).

While limited evidence for genetic discrimination in the context of HD exists (10–12), little is known about how individuals found to have the HD mutation manage the potential for or experiences of genetic discrimination. Insight into the coping strategies used to deal with genetic discrimination can offer approaches for other persons at risk for late-onset disorders. We have conducted this qualitative study to explore how individuals found to have the HD mutation manage the risk and experience of genetic discrimination.

Methods

This study aimed to explore the strategies that individuals use to manage the risk and experience of genetic discrimination. As such, the grounded theory method was appropriate as it is typically used to develop a theory of a process or interaction in response to a phenomenon, 'grounded'

in the views of participants (19). This qualitative research approach is characterized by simultaneous data collection and analysis. The developed theory is based on constant comparison of data with emerging themes from subsequent interviews. Further data collection and sampling is based on the emerging analysis, aimed at maximizing the similarities and differences among the developing patterns. Thus, it is an iterative process of moving between data collection, theorizing/conceptualizing and sampling based on emergent patterns (20).

Recruitment and participants

Asymptomatic individuals found to have the HD mutation were recruited by mailed invitation from three HD clinics across Canada. Asymptomatic status was confirmed with recent neurological assessments at these clinics. No other exclusion criteria were used for study eligibility. Variation across time since testing, age, gender, marital status and education level was sought, which is in accordance with purposive sampling for grounded theory research. Recruitment continued until data emerging from subsequent interviews achieved adequate saturation of the themes (20), which was determined when no new information emerged during the analysis. This study received the approval of relevant research ethics boards. Written and verbally recorded informed consent was obtained from all participants.

Data collection

Thirty-seven individual semi-structured interviews were conducted by telephone (n = 14) and face-to-face (n = 23), digitally recorded and transcribed *verbatim*. Interviews conducted by telephone and face to face did not vary in overall length or quality, consistent with other qualitative research (21, 22). All interview transcripts were reviewed and checked for accuracy.

An interview guide was developed to reflect the research questions. It was based upon literature on genetic discrimination and prior research on the concerns of persons who are at increased risk for HD. Acknowledging the sensitivity and potential bias introduced with the term 'discrimination', the interview guide did not use the word discrimination rather its definition, differential treatment. This term allowed the participants to reflect on their responses to the potential or experience(s) of GD in an open and non-directive way. Participants frequently alluded to GD using terms such as "issue", "ramifications" and "adverse consequences".

The interview topics included experiences with and concern for differential treatment in the family, social, insurance and employment domains, as well as factors influencing the use of particular strategies to manage GD. Some of the questions included: 'what are your experiences in obtaining or keeping life insurance since learning of your test results?' 'Can you tell me what you decided about telling people at work about your test results and how this went?' 'What do you believe would happen if your employer knew of your test results?' and 'How has having predictive testing changed things in your family?' Follow-up probes were used to encourage fuller descriptions and emotion regarding participants' strategies for their concerns and experiences. (The interview guide was continually revised as data collection and analysis continued and the researchers' understanding of the theoretical concepts developed.)

Interviews lasted 65 min on average (range 50–90 min). At the conclusion of each interview, the well-being and need for further support were assessed. Fieldnotes were maintained following each interview to document what the participants spoke about, their behaviour, intonation, emotional responses, the interviewer's (Y. B.) initial impressions on the results and directions for follow up.

Data analysis

A grounded theory approach with constant comparison analysis was used to explore how participants managed the potential for or experience of genetic discrimination. The analysis process included three sequential steps: (i) open coding, (ii) axial coding, and (iii) selective coding. The analysis began by examining the text and identifying descriptive labels for the data. This process fractures the data into the major ideas brought up by the participants. These first-level codes were condensed and conceptual labels (categories) were generated (i.e. take action in advance to avoid GD, ignoring a GD experience). A coding framework was developed to enable identification of recurrent categories discussed by the participants. Following the initial process of taking the data apart, relationships were explored between the categories in the form of causes, consequences and interactions to generate a theoretical model (referred to as axial coding). During this stage, questions and comparisons were made among concepts and new data to facilitate the discovery of patterns and variations among the data (referred to as 'constant comparison'). The final analytic step

of selective coding involved integrating all the categories under a core abstract category or central phenomenon (i.e. managing GD), which connects all categories together to build a theory.

A computer-assisted qualitative data analysis program (NVivo 2) was used to facilitate coding and management of the interview data. Rigour was established by the use of member checking where developing themes and theory were presented to participants for verification throughout the analysis. The coding framework and developing theory was also presented to experts in the field for further validation. Finally, recursive questioning during interviews also contributed to data validation.

Results

Participant demographics

The demographic characteristics of the 37 participants are illustrated in Table 1. These characteristics are similar to previous reports of Canadian adults seeking predictive testing and were found to have the HD mutation (23). In fact, previous studies have reported an excess of females seeking predictive testing for HD (13, 15, 23–25). Thus, this sample appears representative of individuals among the HD population that receive increased risk predictive testing results in Canada. Furthermore, this sample is highly similar to the general population in most demographic variables, although it is less ethnically diverse (97% *vs* 87% European decent) and slightly more educated (89% *vs* 77% high-school graduates) (26, 27).

Table 1. Participants' demographic information

	Participants with HD mutation (n = 37)
	N (%)
Gender	
Female	23 (62.2)
Male	14 (37.8)
Marital status	
Married/common-law	23 (62.2)
Single/separated/divorced/widow	14 (37.8)
Education	
Some college and above	31 (83.3)
Highschool and below	6 (16.2)
Employment	
Employed	26 (70.3)
Unemployed/homemaker	11 (29.7)
Children	
Have children	27 (73.0)
Have no children	10 (27.0)
Time since testing	
0–4 years	9 (24.3)
5–9 years	16 (43.2)
10–14 years	10 (27.0)
15–20 years	2 (5.4)

Dimensions of the GD strategies model

In the course of discussions on GD it became evident that participants attempted to manage both the effect of a GD experience as well as the potential of its occurrence in the future. Participants used these behavioural responses to protect themselves and family from GD and to preserve financial and social opportunities. Although differential treatment may present as positive (advantageous, e.g. increased support) or negative (disadvantageous) treatment (28), strategies were used to manage the effects or potential of a negative GD experience. We noted that participants' behavioural responses or 'strategies' varied along two dimensions: the level of engagement with GD and the nature of the GD experience (actual experience or concern for its potential).

Participants' level of engagement with GD was reflected in the way they dealt with the potential for or experience of GD and varied from high to low levels. Individuals who were highly engaged with GD formed an understanding of it and factored it into their own as well as their families' lives. Engaged participants acknowledged the relevance of GD and directly attended to its potential or experience in an active fashion. In contrast, participants who engaged with GD to a lesser extent did not directly attend to GD and managed its potential or experience in a reserved or limited way. For example, participants did not actively reflect upon the experience nor made strong connections between their experiences and GD (29).

The nature of the GD experience, i.e. whether individuals were managing actual experiences of GD or concerns related to the potential for GD, also influenced the type of strategies used. Those who were concerned about the potential of GD included participants who never experienced GD as well as those that had a GD experience and wanted to manage the future occurrence of another GD event. Participants were concerned about the potential of GD in various contexts, including their workplace, insurance, health care, family and social relationships. There were varying degrees of concern for themselves and their family members. In contrast, participants that had an actual experience of GD responded in ways to manage the effect of that experience. (It is important to note, however, that references to the participants' experiences of GD are based

on their perception of the event that occurred. Thus no distinctions have been drawn between their perceptions and actual experiences of GD.)

Depending on participants' level of engagement with and experience of GD, four discernable strategies to manage GD were reflected in their accounts: "keeping low", minimizing GD, pre-empting GD and confronting GD (See Fig. 1). Although a certain strategy was dominant for each individual in a particular context, strategies varied across time and context and depended on the number of concerns or GD experiences.

"Keeping low"

"Keeping low" was the strategy used to manage GD by individuals who displayed a low level of engagement with GD and were concerned about its potential. This behavioural strategy involved attempts to pass or carry on as though they did not have a stigmatizing identity due to their genetic test results. Overall, 23 participants (62%) described using this strategy.

Keeping private about one's family history or genetic test results was a predominant feature of this strategy. Participants kept private about their risk to varying degrees. Some participants spoke about not "sharing the information" at all or only with a "very limited" group of people. This group typically included family and close friends, described as the "inner circle". Others approached their predictive test results in a protective or "cautious" fashion, concealing their genetic test results more than their family history. Ben (pseudonyms have been used to protect the identity of participants), a participant in his thirties, explained:

> I'm a bit more careful now I think, just I don't really tell many people I've tested positive. Whereas I might have told them that there was this family history of Huntington's, I probably did tell a few friends about that. But I think now that the result was positive I haven't told many people.

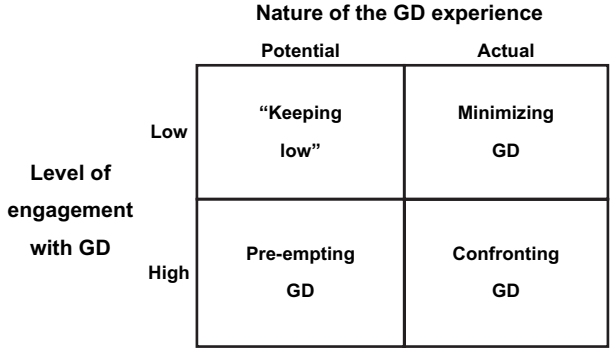

Fig. 1. Strategies to manage genetic discrimination (GD).

Many participants who "kept low" by deliberately keeping their test results private reasoned that such information was "unnecessary" or "none of their [others/employers'] business". The distinction between "having a gene and diagnosis" was a particularly important point for some participants. Prior to diagnosis, they were healthy and did not feel that this information was relevant. Moreover, participants explained that since the disease is the "way it is" with an undefined time of symptom onset they may perceived as 'crying wolf' by disclosing their predictive test results. Wesley, a corporate executive, described his decision to keep private as a "struggle" as he explained:

> I struggled a lot….I thought about if, you know, taking on a new role, taking on a new job which I've just recently done, is not something I should do, it's not something I should tell somebody about, you know, should I tell somebody that I have this condition that in five years time might affect me and all of that and I said, you know what: 'no'.

Those who worried about the possibility that disclosure of their genetic test result may lead to "judgment" did not want to have people wondering about their job performance. Rachel, involved in middle management, did not want to give her co-workers or employers "a chance" to treat her negatively, a sentiment shared by others. Another participant perceived the notion of disclosure as "ludicrous" conveying the strong endorsement for this strategy, which was shared by other participants. In contrast, others spoke of this strategy as a "preference". Some participants employed this strategy in a default fashion, rather than a predetermined plan. These participants suggested that their genetic test results simply "never came up" or "never been an issue".

"Keeping low" was also reflected in actions related to avoiding changes in employment or insurance arrangements. Individuals explained that they stayed in their job in order to avoid the potential loss of insurance benefits, while others did not bother applying for insurance because they were convinced that they would not qualify and thus avoided the probable rejection. Participants also found it important to "keep low" because of the inherent connection between employment and insurance contexts where disclosure in one area may lead to disclosure in the other. Also, laying low was perceived to be

necessary when some participants were concerned about GD in unknown contexts. Hugh, a father of three, explained the nature of his concerns for his children in a variety of contexts:

> I mean… [GD can occur in] any context that they [children] operate in, I suppose, might be potentially one that they could be treated differently in. As I say they go to school and they go to work and they have their friends and social settings and you know potentially even family settings, I suppose…

Some participants also kept low because they perceived their HD risk status as something potentially stigmatizing. A few participants spoke of their HD risk status as "in the closet", a notion typically associated with stigma and shame. Charles, a married father of two, perceived his HD status as potentially stigmatizing for his children when he asserted: "There's enough bias out there based on [one's] religion and race, they [his children] don't need anything else to jump into the picture". Thus he kept "it tight" because of the perceived sensitivity of the information.

Some individuals conceivably used this strategy as a general coping or avoidance mechanism. Upon reflecting on disclosing her HD status to others, Kate, a participant in her twenties, exclaimed: "Oh god…. I would have to explain the whole situation over and over again….it was tough going through it and tough dealing with the answer when I got it". Keeping low may be considered as a strategy to cope with the test results in general.

The level of familiarity and trust with another person or contact was an important factor in determining how individuals kept low. For example, Rachel recognized that she disclosed her HD status to her old boss because he "was a friend" whereas her current boss was not and could not be trusted to refrain from using the information in the "wrong way". Level of familiarity and trust were perceived to be important factors in how participants kept low.

Minimizing GD

Minimizing GD characterized the behavioural strategy of participants who had experienced GD and had low levels of engagement with GD. Typically, participants using this strategy did not directly reflect upon the experience, nor made strong connections between their experiences and GD. Moreover, some participants were ambivalent about whether particular experiences constituted GD. In these circumstances, individuals screened out the incident(s), in effect minimized the GD experience. This strategy of minimizing GD included backing off, avoiding confrontation and disregarding a GD encounter. Overall, 11 participants (30%) discussed using this strategy.

Many participants discussed backing off from a GD situation, essentially not pursing the incident further. Individuals "backed off" from legal proceedings where their test results were used against them, others "backed down" when early retirement was imposed on them due to their test results. Patricia, involved in administration, explained her reaction to her employer's demand for her early retirement to minimize their long-term disability policy costs: "I have to retire at my earliest retirement date so that's the only, you know, thing that has been imposed to me is that so, you know, I think that I have backed down".

Although backing off may be considered as shying away from a situation and avoiding confrontation, some participants perceived the use of this strategy as important in maintaining relationships. For example, Rachel discussed her preference of not expressing her discomfort with her mother's change in communication patterns with her following notification of her genetic test results. She explained, "I don't want to address it with her [mother]….I didn't want to bring it up.… I don't want to make her feel like she's getting any kind of pressure from me". Backing off included resignation and an acceptance of GD. Kate shrugged off the suggestion of re-applying for insurance after being denied when she retorted: "Why go through that if you're going to get denied again." She reconciled the experience by concluding she did not need insurance.

Participants reasoned that minimizing GD allowed them to move on with their lives. Patricia explained this approach: "It will have to, you know, be like water on the duck's back, you just ignore it [GD] and do the best you can with it". Participants also discussed how they "blow off" discriminatory incidents claiming: "It doesn't matter". Beth, a participant in her forties, expressed this sentiment after considering challenging her insurance denial: "Then I thought what's the point, it would just be long, drawn out …it [confronting GD] would be a waste of my time and my effort".

Minimizing GD was also a strategy used by individuals who thought they understood the reason behind the discrimination. Upon reflecting on her experience of a retirement date being imposed on her, Patricia said:

> I understand that because… I don't think that I should be able to use the long term disability

> benefits until I'm sixty-five. I think that would be wasting the long term benefits. I can get my pension next year although it's going to be less than what I would normally get but I can understand why they [employers] wouldn't want me to be on long term disability for a long period of time.

In other instances, participants covered or minimized the significance of their GD experience by constructing GD as "logical" or "just business". Elle, a mother of two, maintained that being charged an additional premium for her insurance "wasn't huge". Participants appeared to minimize the consequences of GD in order to reduce the tension or emotional reaction caused by the experience of GD.

Sometimes participants chose to disregard or defer dealing with their GD experiences in order to focus their attention elsewhere. Danielle, a single mother, discussed going through a difficult period in her life when her boyfriend "dumped" her after she tested positive for HD. Her response to this experience was "whatever" because at the time of this incident she was helping her best friend through a cancer diagnosis and also chose to minimize the importance of their relationship. Thus, individuals may minimize GD when they are distracted or required to manage other, more pressing issues. Similar to "keeping low", minimizing GD may also be considered an avoidant coping response to a difficult or potentially damaging GD event.

Participants also minimized the experience of GD when they found other means to get what they were after. For example, Beth thought "what's the point" of confronting the insurance discrimination since she managed to secure life insurance through a group policy, but admitted, "It would have been maybe different if I hadn't been able to get life insurance, period". Kate, who was denied life insurance, also felt "it's not the biggest deal in the world" since she was covered by her work benefits. In addition, when participants anticipated a discriminatory outcome they readily minimized its occurrence. Kate minimized her insurance rejection but also spoke of expecting her insurance denial beforehand. She said, "Well I was expecting it, you know…At the same token it doesn't feel too good but I had the notion that they would, that it was more of a 95% no than the 5% possibility that it would be a yes."

Pre-empting GD

Pre-empting GD was a behavioural strategy used by participants who were highly engaged with GD and concerned about the potential for GD for themselves and their family. Pre-empting GD involved taking action to evade or "protect" themselves from GD. Individuals cited concern for the lack of legal safeguards for GD in Canada as a reason for this approach. Zara, a participant in her forties, explained: "In Canada there's no real laws yet developed about it [GD]… [so] there's some concern …. Until some laws are in place or something…why take a chance". Overall, 18 participants (49%) described using this strategy.

An important feature of pre-empting GD was taking initiative in an open and direct fashion to reduce the potential for GD. The following measures characterized this strategy: purchasing life insurance prior to predictive testing, educating the public about HD and ensuring one's predictive test results were not listed in their GP's medical files.

A predominant example of pre-empting GD was purchasing insurance before undergoing predictive testing or prior to a family member's official HD diagnosis, a strategy frequently encouraged by genetic professionals. In this way individuals may qualify for insurance based on their family histories so that the principle of good faith may be upheld, since applicants have an obligation to disclose any relevant information at the time of application otherwise they risk having their contract annulled. This strategy was described as "slipping through" the insurance system in order to have a "safety net". Similarly, participants spoke of extending this "safety net" to their children. Wesley, a married father of three, proposed purchasing insurance for his children to "protect" them from GD.

> One of the things I can do for them [children] before they have any [genetic] testing done or anything like that is to perhaps buy them some insurance policies… because once you have the insurance it's easier to keep it…. Just have them [children] do the medicals and so then at least they have something with base protection.

Some participants instructed their children to obtain insurance benefits through their workplace and avoid predictive testing until they've secured insurance.

Educating the public about HD was another method of pre-empting GD. Participants described talking to others about HD in an effort to reverse their perceptions of a general lack of HD awareness in the public, referred to as the "ignorance factor". They took it upon themselves to provide factual information on HD whenever possible. Usually this interaction was less personal because the participants did not explicitly mention details about themselves. In this respect, pre-empting GD was a general

educational campaign about HD and at times included an explanation of the availability and implications of predictive testing. In more intimate environments, educating individual contacts with whom participants shared their test results was a subjective and personal educational process, with the intention of avoiding a specific, personal encounter with GD. Rachel described her form of educating others as "giving" her friends a "little lecture on it [HD]" so as to avoid being "treated differently". In a related approach Wendy, a single woman in her fifties, discussed disclosing her test results "fairly early" in romantic relationships to avoid the potential for "adverse reaction" later.

Participants also took measures to ensure that their predictive test results were not listed in their medical files by instructing their geneticists not to send medical letters to their GPs. Some participants did this to minimize the chance that insurance companies discover their predictive test results if they applied for insurance in the future. Rachel said:

> What concerns me if an insurance company says that they want to look through the medical records and they find this [genetic test result], are they now going to say: 'No, we're not going to cover you'?...I don't want anything on my medical file that says that's a positive result because I believe that insurance companies would treat me differently knowing that even though I'm not symptomatic.

Others were concerned that they could be treated differently by physicians when they are diagnosed with HD in the future. Participants also encouraged others to pre-empt GD. Zara told her nephew "Go back to your doctor and tell him to take it [test results] out of the [medical] file, keep it out of there".

Financial circumstances were also taken into account. Those in higher socio-economic positions were less concerned about pre-empting insurance and employment discrimination. Wesley alluded to this after he discussed his plan to purchase life insurance for his children:

> But again financially I'm not too worried because unless we have a huge stock market wreck or a big crash or something, I can help them [children], you know, if they get into a situation where, you know, this affects their ability to get insurance or whatever, you know, I can help them.

Confronting GD

Unlike highly engaged participants who were concerned about the potential for GD, those who were highly engaged with GD and had an encounter with GD confronted it head on, resisting or challenging the GD experience. Confronting GD was characterized by a spectrum of approaches including challenging the perpetrator, seeking advice and refuting the basis for discrimination. Overall, 10 participants (27%) discussed using this strategy.

In responding to differential treatment, such as judgemental comments participants, discussed confronting their perpetrators by "making others listen" and telling others that they "don't want to be treated that way". An experience with GD seemed to empower some to confront further discriminatory experiences. Whistle blowing became a strategy of choice for those with multiple GD experiences. Michelle, a health-care professional, considered her HD test result as an "ace card" for exposing GD. Her boss requested access to her medical files for surveillance purposes after discovering her genetic test results. Although Michelle refused this request, she believed she retained the upper hand:

> I hold the ace card basically because, if they, if I feel I am being shafted in anyway I can pull out the ace card and say prejudice....If I apply for a position and I feel that, you know, they've declined me because of Huntington's...if I feel that in any way I would have no qualms about going to the Times columnist and going hey you know...this is what's happened.

Participants also sought the advice of legal experts, protection of unions and support of friends and trusted health-care providers in confronting GD. Oliver, a participant involved in public transit who had been recently fired at the time of the interview, sought assistance from his union in dealing with the conspiracy he perceived occurred among his physicians and a driving instructor, which culminated in his dismissal. Others who wanted to confront uncomfortable behavioural and communication changes in their family related to GD, sought the support of health-care professionals, such as psychologists. After learning of his test results, Wesley recalled: "She [wife] began to evaluate me through a different lens and she was seeing changes in me that I wasn't seeing". He worried that this treatment impacted their relationship. He encouraged his partner to attend joint counselling sessions "for the sake of the relationship".

Some participants complained of being "shunned" or were treated as though they had a "contagious disease". When they faced a discriminatory situation some individuals attempted to refute the basis for the differential treatment.

They would attempt to explain the distinction between having a gene mutation and having a disease while others would explain to others that they are not currently sick. Beth described her friends' treatment of her after she told them of her genetic test results:

> At first they treated me like I was made of glass, like I was going to break and that lasted for about a month.… [They were] just very-very careful in what they said and how they said it. I mean you could tell the effort was there but they were being very-very careful and everything they said and everything they did so it wouldn't upset me. And that just drove me nuts. And I just looked at them and I said, 'I'm not sick' 'I'm not dying' I said, 'Sure I have this thing but I'm fine'.

Individuals' tolerance for ambiguous or awkward situations determined how they confronted GD. Beth discussed her lack of tolerance for the "shunning" and thus frequently confronted individuals that "have a problem" with her. These personality traits in addition to being generally "strong and stubborn" individuals were perceived to be important factors in how participants confronted GD experiences.

Discussion

The results of this study suggest four broad strategies are used to deal with genetic discrimination: "keeping low", minimizing, pre-empting and confronting genetic discrimination. This typology is presumed to be specific for asymptomatic individuals coping with a potentially discreditable identity as a consequence of being at increased risk for a late onset genetic disease. Given the recent attention surrounding genetic discrimination (30–32), learning how individuals deal with real or potential genetic discrimination is of importance to genetics professionals in assisting individuals effectively mange these issues. Thus, these strategies may provide a framework for understanding how other individuals manage genetic discrimination for other genetic diseases for which predictive testing is available. These include breast and colons cancers, and other neurological and cardiovascular conditions such as spinocerebellar ataxias, myotonic and muscular dystrophies, Alzheimer's disease, thrombophilia, hypertrophic cardiomyopathy and Marfan Syndrome. These findings may prove particularly relevant for cancer genetics settings where calls for new approaches to address genetic discrimination during genetic counselling have been recommended (33).

Research attention has recently focused on the stigmatization and discrimination against asymptomatic individuals at risk for various genetic diseases (10, 30–32). The stigma-related coping literature provides a basis for understanding the results of this study. Stigma can be a source of stress for stigmatized individuals (34) and to cope with it, individuals employ strategies which are aimed at controlling and modifying the situation by using psychological, social, behavioural, economic or educational approaches (35). Similarly, individuals found to have the HD mutation use predominantly behavioural strategies to manage the experience of GD and to control its potential. Although our typology attempts to group responses into clearly defined categories, there can be overlap between categories, especially in different contexts and situations. Consequently, different strategies may be employed by any one person at different times and across various contexts. Individuals' use of different strategies depended on their level of engagement with GD and on the nature of the discrimination experience (actual or concern for the potential of GD).

The conceptualization of engagement as an underlying dimension in explaining how individuals manage the risk and experience of GD is supported by recent studies in relation to coping with stigma-related stress and genetic risk for HD (36–38). Responding to stigma always involves cognitive appraisals about the seriousness and relevance of the threat (36). Moreover, the perception of threat is likely to occur only among stigmatized people who self-identify with the stigma (37). Our findings support these models since a GD strategy is employed as a consequence of the individual's perception that the threat of GD is relevant to them and that they have something personal at stake. In other words, one must be engaged with GD in order to perceive GD as a threat and consequently respond to it. Moreover, the nature of one's response to GD will depend on their level of engagement with GD. Although engagement with GD is likely represented as a continuum, dichotomizing engagement was nonetheless helpful to illustrate the different strategies individuals used to manage GD.

Our results indicate that individuals who engaged with GD to a lesser extent adopted strategies of non-disclosure of their genetic test results in potentially stigmatizing situations and stayed in unsatisfying jobs because of concerns related to having the HD mutation, which is consistent with previous research (32, 39–41). The participants in our study divided their world

into a large group to whom they tell nothing (''keeping low'') and a small group (''the inner circle'') who were informed of their genetic status. Medical practitioners often recommend this type of information management by instructing patients to take caution when and with whom to discuss their test results. The effectiveness of such a strategy may be called into question because some stigma theorists speculate that felt stigma (fear of discrimination) typically proves more disruptive than enacted stigma (actual discrimination) (40). In fact, the use of secrecy to manage social stigma has been linked to emotional distress among caregivers of people living with AIDS (41) and thus it is plausible that individuals who ''keep low'' may experience distress. The pre-test counselling process and informed consent includes consideration of the potential implications and negative effects of having predictive testing, which could be considered an attempt to encourage greater engagement with GD. However, given the potential distress and risk of exacerbating felt stigma inherent in this process attempts should be made to temper these discussions. Given the emphasis of ''keeping low'' as a choice strategy for those at risk for GD, further research is warranted in exploring the effectiveness of using this strategy as well as its long-term impacts on emotional well-being.

Low levels of engagement with GD coupled with an actual experience of GD resulted in the use of strategies focused on minimizing GD among participants in our study. This approach has been previously described as a disengaged response to discrimination in which participants' choose not to reflect upon or discuss the incident (37, 42). Some participants were ambivalent about whether a particular reaction constituted discrimination. Avoidance, acceptance and minimization, elements of this strategy, have also been previously reported to be associated with disengaged coping responses to discrimination (43, 44). Avoidance, characterized as withdrawal, resonates with the participants' desire to avoid confrontation following a GD encounter. Moreover, those who used this approach minimally engaged with GD. In fact, some research has suggested that minimal engagement is a choice strategy to cope with discrimination since individuals are able to successfully reduce psychological distress and, thus maintain emotional equilibrium without taxing their coping resources (44, 45). Conversely, other evidence suggests that disengaged coping responses to stigma are less adaptive strategies to cope with stigma-related stress and may lead to adverse consequences. For example, previous discrimination research has demonstrated that African Americans who accepted unfair treatment were more hypertensive than those that took some action (46). Moreover, African Americans who asserted that they did not experience racial discrimination were more likely to be hypertensive (46). As emerging evidence suggests, individuals that chose low engagement strategies may experience less distress yet perhaps at the expense of their physical well-being. Further research would be required to establish correlates between low engagement strategies and psychological or physical outcomes.

In contrast, those that engaged with genetic discrimination to a greater degree responded to its potential by pre-empting it. Adapting one's social interaction strategies included behaving in a socially skilful manner in the face of prejudice. Participants' measures of purchasing insurance policies before genetic testing and keeping test results out of their GP's medical files may be considered as dimensions of pre-empting the risk of genetic discrimination and attempting to achieve their goals despite their genetic difference. Educating the public, an important measure of pre-empting genetic discrimination, was evident as participants embraced opportunities to inform others about HD and in doing so attempted to change other's negative perceptions of HD. Similar approaches have been adopted by others. For example, 51% of respondents on a survey on coping with Marfan's disease endorsed the use of education as a strategy to cope with disease-related stigma (32). The use of preemptive strategies may be likened to 'passing', an important response described by Goffman (1963) in which individuals act as if they have a less stigmatic identity or even a normal one (47). Although the relative effectiveness of pre-empting GD is unknown, our results suggest that this strategy may be employed in a range of contexts and circumstances.

Resistance has been a described as a response to stigma in which participants speak out or challenge rules or the stigma (48). Similar to our results, some individuals experiencing GD confronted the incident by challenging the person or institution responsible, seeking professional advice and refuting the basis for GD. These engaged strategies are characterized by a 'fight' motivation (35) and an attempt to change these circumstances. Our findings suggest that participants who confronted GD did so in certain contexts (i.e. social and family settings) and were largely individuals with a low tolerance for ambiguity.

There are several caveats in the interpretation of this study. First, participants were recruited

from a larger observational study of individuals who underwent predictive testing for HD. This recruitment strategy may have biased the findings because the participants may be considered a self-selected group (49). In addition, our study primarily explored behavioural responses to the potential or experience of GD. The array of responses to stigma is vast, including emotional, cognitive and physiological responses, which can occur both voluntarily and involuntarily. Consideration of the diversity of coping responses may be necessary to gain a complete understanding of the consequences of GD. The interpretations and typology is thus tentative and we are unable to immediately generalize to a larger population. Additional research is warranted to explore the predictors and outcomes of using these strategies in a more broadly representative sample. These insights would be helpful to predict the variation in approaches and reactions to GD among individuals found to have the HD mutation.

The results of this study have implications for the care of asymptomatic individuals living with a positive test result for HD. Genetic discrimination can be detrimental to one's well-being. Genetics professionals can assist their patients to anticipate struggles and encourage the use of engaged strategies to help them manage GD. Thus in the context of genetic counselling for HD predictive testing, it is imperative to explore patients' experiences with stigma or GD and assess individuals' resources to cope with GD. Furthermore, clinicians should mobilize effective engaged strategies and refer individuals who are struggling with issues of discrimination for additional support or legal counsel. It is through insights from stigmatized individuals that we may learn how to help others cope with potentially discreditable identities as a consequence of testing positive for a late onset genetic disease.

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