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In their own words: Reports of stigma and genetic discrimination by people at risk for Huntington disease in the International RESPOND-HD study

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Abstract

Genetic discrimination may be experienced in the day-to-day lives of people at risk for Huntington Disease (HD), encompassing occurrences in the workplace, when seeking insurance, within social relationships, and during other daily encounters. At-risk individuals who have tested either positive or negative for the genetic expansion that causes HD, as well as at-risk persons with a 50% chance for developing the disorder but have not had DNA testing completed the International RESPOND-HD (I-RESPOND-HD) survey. One of the study's purposes was to examine perceptions of genetic stigmatization and discrimination. A total of 412 out of 433 participants provided narrative comments, and 191 provided related codable narrative data. The core theme, Information Control, refers to organizational policies and interpersonal actions. This theme was found in narrative comments describing genetic discrimination perceptions across employment, insurance, social, and other situations. These reports were elaborated with five themes: What they encountered, What they felt, What others did, What they did, and What happened. Although many perceptions were coded as hurtful, this was not true in all instances. Findings document that reports of genetic discrimination are highly individual, and both policy as well as interpersonal factors contribute to the outcome of potentially discriminating events.

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INTRODUCTION

Huntington disease (HD) is an autosomal dominant neurodegenerative condition, with a prevalence of approximately 4–7/100,000 (Online Mendelian Inheritance in Man). Symptoms encompass progressive loss of cognitive, behavioral, and motor functions, with an estimated lifespan of 10–25 years after motor diagnosis (LoGiudice and Hassett, 2005). Neuropsychological testing of people who possess the HD gene expansion, but who do not manifest motor symptoms, suggests that detectable changes in cognitive function occur 1–2 decades prior to clinical HD diagnosis. In addition, psychiatric disturbances, olfaction, striatal volumes, and subtle motor signs are also detectable prior to clinical motor diagnosis (Paulsen et al., 2008). Perceptions of stigma or discrimination may be influenced by these subtle neurologic changes prior to clinical diagnosis. However, perceptions of stigma or discrimination by individuals with prodromal HD or with a family history of HD may influence decisions they make in their day to day lives. Although advances in early biological and refined clinical markers of disease have burgeoned in the past decade, little is known about the stresses of living with the knowledge that one will someday develop HD (Beglinger et al., 2008).

Genetic discrimination refers to adverse treatment based solely on the genotype or family history of people without symptoms of a disease (Rothstein and Anderlik, 2001). A related concept is stigma, which refers to the co-occurrence of components that include labeling, stereotyping, separation, and status loss (Link and Phelan, 2001). A central feature of stigma is the perception of an attribute of the stigmatized person that conveys a devalued social identity within a particular social context (Crocker et al., 1998a). Stigma is a phenomenon that is difficult to disentangle from interrelated perceptions and experiences of discrimination events by people at risk for HD, and both family history and genetic testing may be the basis for either stigma as well as discrimination (Biser, 2004).

The central feature of stigma is the perception of an attribute of the stigmatized person that conveys a devalued identity within a particular social context. There are many different attributes to stigma, including objectification, devaluation of the stigmatized person, or demeaning behaviors directed towards the individual. Stigma may involve minor daily hassles, such as being ignored in a group, or it may involve discrimination in the form of being denied job opportunities, education, or insurance benefits (Crocker et al., 1998b). Stigma is an especially difficult stressor on individuals because it can become a self-identifying “master status” that encompasses qualities which become a core part of the identity of the individual affected (Goffman, 1963). Thus, the perception of stigma may not be distinguished from discrimination in the minds of people who experience it.

In this study, perceptions of discrimination were not limited to situations governed by laws, regulations, or policies, but also included those that may result in prejudice, loss of privileges, or personal hardships. Thus, the definition of genetic discrimination in this study is a broad one that encompasses all perceptions of adverse treatment based solely on genetic information. Understanding the influence of possible or actual genetic discrimination in the day-to-day lives of people at risk for HD has received little attention until recently. A review of interviews of such individuals noted that genetic discrimination was reported with insurance, in the workplace, and within social relationships (Penziner et al., 2008). A larger study of people at risk for HD focusing on individuals who, initially, had not pursued

genetic testing found that those who subsequently did pursue genetic testing were fearful of losing health insurance, in some cases actually paid out-of-pocket for the test in an effort to conceal their genetic risk from their insurers and/or employers (Oster et al., 2008). A prior study of Canadians at risk for HD found the response to the experience of (or potential for) genetic discrimination depended on the person's examination of both the situation and ways in which people form meaningful interpretations of genetic discrimination and its risk and consequences (Bombard et al., 2008). Strategies for dealing with the fear (or actual experiences) of discrimination varied according to both the individual's engagement with genetic discrimination and the nature of the experience (Bombard et al., 2007). A large Australian study examined reports of genetic discrimination by 951 people who had presented to clinical genetics services for genetic testing and were asymptomatic at the time of the survey. Findings suggested adverse treatment by 10% of the sample, with those having neurodegenerative conditions being more likely to report incidents of discrimination or social stigma across multiple domains (Taylor et al., 2008). Taken together, these reports document a pattern of concern regarding the actual or perceived threat of genetic discrimination that extends across employment, insurance, social, and other situations within the population of people at risk for HD.

Methodological limitations of studying discrimination include the unlikely prospect of having discrimination confirmed by the perpetrator. Thus, with the exception of one study (Barlow-Stewart et al., 2009), reports of genetic discrimination consist of self-reported data elicited through surveys and interviews, without corroborating second-party accounts. Barlow-Stewart et al. (2009) report that 93 asymptomatic individuals in Australia reported incidents of alleged negative treatment. Fourteen of these cases proceeded through the verification process, and of these, three were not verified by insurers. Despite difficulties in validating reports of stigmatizing or discrimination experiences, concern about these issues remains among people at risk for HD, health care providers, and policy makers. The purpose of this report is to examine perceptions, of genetic discrimination (i.e., unfair treatment by others) as reported by individuals with a family history of HD.

MATERIALS AND METHODS

This research is a component of a larger study (Paulsen et al., 2005) whose purpose was to examine the reports of adverse effects among people who underwent genetic testing for the HD gene mutation and to determine the factors that may influence any adverse effects of genetic information in terms of regulatory systems. A descriptive qualitative design was used to examine reports of genetic discrimination by people with a family history of HD, as well as those who have had a predictive test for HD, regardless of whether they have the gene expansion. All study participants were considered healthy and did not meet criteria for manifest disease. Reports of stigma or genetic discrimination were elicited as a component of a larger study in which people at risk for HD in the United States, Canada, and Australia completed the International RESPOND-HD survey to describe their perceptions of HD related stigma or discrimination (Erwin et al., 2009). The survey contained both closed- and open-ended response items. In the open-ended sections of the survey, participants were asked to describe incidents of genetic discrimination or to provide further information about the events. This paper reports findings from this narrative information.

Recruitment and Data Collection

The participants for the I-RESPOND-HD survey were concurrently enrolled in either the PREDICT-HD (Paulsen et al., 2006) or the PHAROS (Huntington Study Group, 2006) studies or persons attending the 2007 Huntington's Disease Society of America Convention in Oklahoma City who met inclusion criteria. The PREDICT-HD Study is an ongoing multinational observational research study that evaluates genetically at-risk individuals who,

prior to enrollment, have undergone elective DNA analyses for the HD gene expansion and who were not clinically diagnosed with motor symptoms of HD at the time of their enrollment. The purpose of the PREDICT-HD study was to identify the primary emerging biological and clinical precursors of HD and establish what factors influence the age of symptomatic HD onset (Paulsen et al., 2001). The study includes men and women, 18 years of age or older. The PHAROS Study (Huntington Study Group, 2006) was a multinational observational research study that evaluated genetically at-risk individuals who had previously not undergone elective DNA analyses for the HD gene mutation, who were not clinically diagnosed with symptoms of HD at the time of their enrollment, and who did not wish to know their genetic test results. The study aimed to identify the emerging clinical precursors of early symptomatic HD onset in a reliable and gene-specific manner (Huntington Study Group, 2006).

I-RESPOND-HD participants were recruited by site coordinators at three Australian, two Canadian, and ten US PREDICT-HD or PHAROS research sites, as well as the Huntington's Disease Society of America annual meeting (Erwin et al., 2009). The I-RESPOND-HD survey was conducted from January 2007 to December 2008 in compliance with the Code of Ethics of the World Medical Association (Declaration of Helsinki), and Institutional Review Board (or equivalent) approval was obtained at each site.

Survey Instrument

A full description of the I-RESPOND-HD questionnaire is reported elsewhere (Erwin et al., 2009). In brief, the questionnaire domains and items within each domain were developed through examining the literature and analyzing the interviews conducted with people at risk for HD. The survey included sections addressing stigma and genetic discrimination based on family history, genetic test results, and specific incidents of insurance, employment, family, and social issues (Erwin et al., 2009). Two additional blank pages were included at the end of the survey to allow for continuation of narrative responses, if needed. Narrative comments ranged from short phrases to extensive descriptions.

The I-RESPOND-HD survey development included piloting of the survey. This measure used the terms "differential treatment," "treated unfairly," and "discrimination," throughout the survey in an attempt to minimize response bias (Treloar et al., 2004) while clearly communicating the intent of survey items. Each section contained both closed- and open-ended response formats in order to capture descriptive and contextual data through narrative comments, as well as an account of such occurrences and the contributions that others made to the described experiences.

Survey items were developed, in part, from interviews. These interviews yielded the major domains for the survey that reflect perceptions of employment, insurance, societal, and family experiences, and confirmed feasibility of eliciting data on genetic discrimination from people at risk for HD. Four HD experts reviewed the survey for comprehensiveness and clarity. Six at-risk individuals completed cognitive interviews to confirm survey usability (Drennan, 2003).

Data Analysis

All narrative comments were extracted from the surveys and content analysis methods were used to code narrative data by between three to six members of the research team (Knafl & Webster, 1988; Sandelowski, 2000). All data were coded into the software data management program (NVivo, 2008) for use in descriptive qualitative analysis. These data were first coded into 17 descriptive categories according to descriptive analysis procedures (Knafl and Webster, 1988). Differences among coding determinations were discussed until there was

100% agreement. Categories were then refined, reconfigured, and sorted by two of the team members who coded the data and a third team member who had coded the data into themes. These three team members conducted further analysis of all data within these categories to derive an overarching core theme. The core theme and themes were not confined to the questionnaire sections from which they originated; instead, they represented findings derived from the narratives of the situations in everyday language from data that cut across the survey as a whole. Themes reflected the ways in which the experiences unfolded throughout the narrative comments. Members of the analytic team who conducted the original coding reviewed and agreed that the subsequent analysis was an accurate accounting of the events and their meanings (Sandelowski, 2000). Although each participant did not provide a longitudinal narrative of all aspects of the incident, the overall data provided a rich description of the participants' perceptions of events of stigma and discrimination and how these evolved in the participants' lives.

RESULTS

Sample

Four hundred thirty-three participants completed the survey (Table I) (Erwin et al., 2009). The majority of the respondents were from the United States, and there was higher overall percentage of female respondents across all countries. Participants were at-risk individuals who had tested either positive or negative for the genetic expansion that causes HD and at-risk family members who have a 50% risk for developing the disorder but had not had DNA testing. Four hundred twelve provided narrative comments (see Table II). Selected demographic data for respondents of quotes to illustrate each theme are provided in Table III.

Core Theme: Information Control

Information Control was identified as the core theme. "Information Control" reflects privacy and confidentiality concerns, implicating control of an individual's genetic information. Privacy concerns address the collection of, or access to, individually identifiable genetic data. Confidentiality concerns involve the disclosure of data previously entrusted to another. Both genetic related stigma and discrimination arose from the loss of control over how genetic information was accessed or used.

Regardless of the perception of the type of experience being described, Information Control was embedded within the descriptions of individual participants' experiences. This core theme reflects situational and interpersonal influences within the occurrence, the response by the participant to the occurrence, and (in some cases) resolution of potential genetic discrimination. Just as there is no one best way of preventing or coping with genetic discrimination that will fit all people and circumstances, there was no single strategy that participants used in their attempts to achieve the results they sought. Life experiences, naiveté, bad advice, good advice, and the notion of planning were all part of the overall set of narrative descriptions. Throughout the narrative comments, individual characteristics of the respondents and the people involved in the discrimination influenced these reports. Although genetic discrimination is defined as adverse treatment of a person, seldom was this described as a solitary event. Instead, it was often described within the context of actions and intentions of others. Strikingly, the instances of negative interpersonal interactions ranged from overtly offensive to covert and subtle, and in some cases, perceptions of these experiences included deliberate and persistent advocacy on the part of others involved in the occurrence.

Five themes were identified as components of the personal experiences that gave meaning to individual events under the core theme of Information Control: What They Encountered, What They Felt, What Others Did, What They Did, and What Happened. Each theme is presented here with examples of data illustrating the theme (Table IV).

Theme: What They Encountered

Participants (Table III) reported a range of ways in which they encountered obstacles in gaining access to goods and services that are normally available to healthy individuals.

Institutional Factors—Respondents encountered organizational policies that eliminated them from opportunities; these were mainly with regard to insurance. For example, this participant's spouse provided the information without the person's consent: *because of my pre-existing condition...[health] insurance would cost more if I could get insurance at all* (1). Others noted that they were requested to give genetic information they did not want to reveal. Despite their desires, some participants provided the information and asked that it not be disclosed or used in some way, but this did not happen. The result of policies, i.e., practice guidelines that may be written or are understood to be standard operating procedures, that requested genetic status was that the participant lost control over how their information was disseminated or used. In some cases, physicians put genetic information in medical records against the request of the patient. For example, one participant noted: *I told my prior MD that I had tested positive for HD. After switching MDs I found out that he had put these results in my medical records after I had told him not to* (2). Another stated: I *was denied life insurance. My doctor, who was a personal friend of mine, put my family history and gene status in my medical file when I specifically asked him not to. Many years later I was getting life insurance and was denied when they saw the reference in my file. I should have never told my doctor* (3).

Interpersonal Factors—Regardless of institutional policies, respondents encountered various reactions in interpersonal settings that resulted in discrimination. The first example of this was when others were prying into employment or insurance information, and individuals felt pressured to reveal information they otherwise would not. This pressure was in the form of false support or promises of compensation, financial or emotional, in return for revealing the information. Individuals referred mostly to insurance and to employment, especially during the job interview phase. One participant offered an example in which colleagues provided information to the manager without the participant's permission: *Colleagues feigned support whilst feeding information to manager* (4).

The second type of interpersonal factor was "stonewalling," which refers to stalling or making fairly neutral remarks that seem to cover for a discriminatory intent, at least as perceived by the participants. Most examples of this factor referred to employment. One participant volunteered test results and family history to an employer without being asked: *I had shared with people at work when I found out about the test results. Most of them, counselors, I thought it was a safe environment. I had been watched and had been told I was having problems getting along with others. I then was told that I did no longer* [sic] *have employment* (5). For some, these interpersonal factors led to loss of employment when there was not evident cause for the termination other than HD risk, as perceived by the participant. Another participant also had volunteered her family history of HD to the employer without being asked: *After completing teaching in-service training for staff on HD, my position was terminated… kept me employed until I did find another job* (6).

Interpersonal factors were also cited when participants related poor treatment by family members. This group of people perceived experiencing attitudes of family members or

actions that were not limited to the contexts of insurance or employment. For example, family members requested the individual not have any future contact with the family, they abandoned the person, blamed them for ills of other family members, or gave unwanted advice about personal decisions such as family planning, life planning, financial planning, or companionship needs. A participant had volunteered HD family history and genetic test results to family and employer without being asked: *Family disowned me… Called my employer, coworkers, friends… they're all just angry that I tested* (7).

Poor treatment by others outside the family was also described. Participants felt they were seen as vulnerable by others and reported experiencing ill treatment such as bullying, abuse, ostracism, or cruelty from others who may be peers, strangers, or even friends. Another volunteered the HD family history to friends without being asked: *All my friends stopped calling and being friendly after results… too busy… most no excuse* (8).

Theme: What They Felt

Respondents described their emotional reactions to perceptions of experiences of discrimination. Some individuals felt overwhelmed by the experience of stigmatization or discrimination while others received the event as a call to action.

One reaction was a sense of injustice. Feelings included impotence in which some participants sought out retribution, but no retribution process was available to some persons. For these participants, seeking justice could happen if only they had the resources. A participant reported that a fellow employee found out by asking family members, and then turned the participant in for being unsafe at work. The participant reported he was subsequently fired and lost of some insurance coverage: *I don't have the money to challenge them legally… it takes money to hire a lawyer and to fight it and to set the precedence* (9).

For others, the emotions were a sense of futility, in which there was no hope for justice due to inherent power or hierarchical differences. This participant noted that her HD status was in the medical file: *I didn't complain because he's a doctor* (10).

Other emotions reflected fear or a sense of vulnerability such that some didn't dare step out from the safety of silence. This was a more debilitating type of fear in which they didn't have the will or the desire to fight. Feelings of regret were coupled with feelings of being wronged. One participant referred to a time after disclosure of a sibling's HD illness to friends at work: *I was afraid they would fire me for something else. I never felt comfortable/ secure in my job after that* (11).

For some, perceived genetic discrimination led to a more direct response, or a stirring to action. Some expressed anger, while others felt empowered. One person, noted: *I went to the CEO & made it public knowledge* (4). Another, who describes herself as an HD advocate, reported frequently telling people of a family history of HD: *Angry… no one being educated* (12), while another person offered: … *just knowing helps prepare for the future* (13). This person reported being watched for symptoms after undergoing genetic testing, but he kept the results a secret.

Theme: What Others Did

Not all respondents were left to their own resources when genetic discrimination occurred. Although there were fewer comments in this category, for some, other people provided valuable help and direction to remedy the situation.

Some respondents reported the advocacy of others. In some cases others were describing as having intervened to assure that the discrimination would not occur. In one instance, the

person was told by the insurance agent not to ask for a certain high amount of life insurance. This participant reported volunteering her HD family history to the insurance agent: *Insurance agent advised me to only apply for a certain limit of insurance so we didn't have to give a blood sample* (14). In other cases, perseverance and action by others reversed a policy that could have led to discrimination. In one case, the participant reported telling a blood bank about her predictive test and being told she could not donate blood anymore: *The doctor rang & told me. I told him I wanted the decision in writing and I would take it to the geneticists, HD counsellor, HD association & media (if necessary). The doctor said he'd try again. I received a letter saying I could donate (blood) again* (15).

Theme: What They Did Themselves

Respondents described finding strength in themselves and a turning inward. One example is relying on one's personal policy for making decisions. For example, one participant described herself as presymptomatic and reported a situation when she perceived that discrimination came from others who stared at her mother when they traveled, because of her mother's jerky movements: *Confront the situation with a positive attitude and put in my best* (16). Some respondents challenged others, such as a participant who volunteered genetic test results and family history to an employer without being asked: *I wrote a letter of complaint to the GP, employer* (17). They also changed the situation. They persisted until they got the results they wanted. Another person avoided the potential for an unpleasant situation. *I went to an ENT about a lump in my neck. He treated me like I wasn't worth the trouble and lectured me for having kids even though I had no knowledge of my family history until my kids were grown. I didn't complain because he's a doctor. I just don't go to anyone but my neurologist now.* (10).

It is clear that direct actions are not the remedy for every situation or for every person. For some, guarding their privacy was the preferred response. By keeping information to themselves, they were able to maintain their image of how other people saw them. Some participants described decisions to avoid risk for genetic discrimination. One example is: *I would like to comment that I have had no incidents of discrimination because I do not tell most people about my genetic test or family history. It has worked out very well* (18). Another respondent wrote: *Of course I tested anonymously so my company and insurance companies don't know. Information is power. With HD we have to think and act proactively* (9). One participant noted that she was originally denied life insurance when she was asked for, and provided, her family history: *I transferred to the US to work, luckily they have non-discriminatory life insurance, got some and kept it* (19).

Others dissembled, using subterfuge and deception as a counter-strategy to deal with perceptions of genetic discrimination. I obtained long-term care, disability, health, life insurance years before I might have ordinarily. I didn't mention HD to my doctor. My mother's cause of death is listed as pneumonia… so when asked, that's what I put… that is how my dear mother benefited me (20). Still others found their fear of the consequences prohibited them from taking any actions. I haven't been brave enough to ask for insurance or get advice from anyone (21).

Theme: What Happened

Several respondents concluded their narrative comments in the survey by describing the outcome of a situation that they perceived had led to discrimination. For some people, they did not reach a satisfactory resolution. For example, *We were accessing funds to build a house. The business suggested a Life (insurance) policy to cover the mortgage in the event of death. Decision was made on the genetic testing data, which I provided. It didn't seem to matter about our family history having very late onset HD. I was refused life insurance,*

which was required for a house mortgage in the event of my or my husband's death. Due to lack of funds, I was unable to pursue legal action (21).

Not all resolutions were disappointing. A participant reported a situation where she provided gene test results and HD family history to an insurance company when asked to do so: *I had a court case for a car accident. HD, as an issue, was raised but deemed irrelevant by the Judge. In court it showed me to be a responsible person acting maturely by taking the genetic testing. The opposing Insurance company wanted to blame me, and for me to be responsible for the car accident because I had the HD gene. I was awarded 100% of the claim* (22). And another person provided family history information to an insurer when asked to do so: *I signed up for long-term care through my employer and they (the insurance company) did not want to approve me. When (they) repeatedly dragged their feet, I challenged them as to whether or not their excessive delay had anything to do with having a family history of HD in my files—they never denied that, but eventually did approve coverage* (23).

DISCUSSION

This study provides the first report of perceptions genetic stigma and discrimination in a large data set of people at risk for HD across three countries. Many participants offered qualitative narrative comments on this survey, possibly reflecting a need to share concerns about this topic. This observation reinforces the importance of qualitative research methods for more fully understanding this highly personal topic. This study provided analysis of data from open narratives in which respondents provided contextual information regarding not only their perceptions about genetic discrimination and stigma, but also their response to it.

Findings emphasize that revealing test results or genetic family history to an employer continues to be a situation for which people at risk for HD perceive that they face the potential for genetic discrimination. Bombard et al. (2009) report perceptions of employment discrimination by 6.9% of a sample of Canadians at risk for HD. As noted by Penziner et al. (2008) in their interviews of 15 at-risk people, although most participants had revealed results to an employer, they also said they would not do that again. In the present study, narrative comments document perceptions of the insidious nature of experiences in which seemingly supportive or ordinary social interactions could not be either directly linked to nor disentangled from subsequent events in which the at-risk person had no other explanation for loss of employment status. These insights are important, as they serve to raise questions regarding the dynamics within which the at-risk person, co-workers, and employers navigate workplace relationships that would influence one's abilities to sustain one's employment. Influences of the public's perception of HD, and concern for employers' financial or workplace risk when employing a person with genetic risk, are topics for further investigation.

The analysis of responses portrays societal factors in addition to personal coping styles when people at risk for HD are faced with the potential for, or perception of the occurrence of genetic discrimination. A surprising finding was the nature and extent of help that some people received. Sadly, others received no help and were then left with only their personal resources, which varied widely from feeling that any response was futile to assertively pushing back in an attempt to right the perceived wrong. Findings from this study report tragic as well as triumphant stories and add to the growing body of evidence that stigma and genetic discrimination are perceived to occur, may have differing outcomes, but are not experienced by everyone.

One area of particular concern is the privacy and confidentiality of health information and health records. The legal structures governing the privacy and confidentiality of health information do not endorse absolute confidentiality of health records due to countervailing public policy concerns (Terry and Francis, 2007). Although neither privacy nor confidentiality provides an absolute barrier to access of genetic information, some measure of control remains within the grasp of individuals wishing to self-define their personal and social relationships. However, this control is limited, and the inability of individuals to fully control the access to (and disclosure of) their genetic information provides ample opportunity for institutions and individuals to learn (or seek to learn) about genetic information.

The overarching theme in this study, Information Control, also concerns the use of genetic information. Once genetic information was accessed by others, it could be used either appropriately or inappropriately to deny positions, opportunities, or other advantages. Medical information, including genetic information, may be given to others and used appropriately to make decisions, such as hiring decisions that are consistent with the Americans with Disabilities Act or for other purposes (Miller, 2000). Genetic information may also be used inappropriately to deny opportunities for which an asymptomatic individual is otherwise qualified (Rothstein and Anderlik, 2001).

When stigmatization and genetic discrimination were perceived to have occurred, these occurred across circumstances for which there may be laws addressing genetic discrimination, or in social or family settings in which there are no formal antidiscrimination processes. Individual stories provide insight into the burdens of living at risk for genetic illness, and show many respondents keeping their genetic status private, even from their family. The reports from our participants reinforce the notion that being at risk for a neurological disease can have a strong stigmatizing effect. As the world's population ages and an increasing number of individuals anticipate dementia of any etiology, the knowledge of this frequency of stigma and discrimination may inform public policy in addition to private decision making. Some participants reported encountering regulatory issues regarding insurance or employment. Even in these examples, however, many comments addressed behaviors by people that were both beneficial and harmful. These reports further clarify that regulatory changes alone won't necessarily lead to diminished genetic discrimination unless personal intentions and behaviors also change.

Progress to reduce stigma and genetic discrimination will be hindered if efforts are limited to policies and the response capacity of individuals without attention to the societal context within which these events occur. The need for better understanding of HD by health care providers and the public is reported as being the most pressing problem faced by HD families in the US and UK (Williams et al., 2009). As more illnesses with a substantial genetic component are revealed, the need for education, empathy, and tolerance for genetic risk will increase as resources and remedy for genetic discrimination. Efforts to educate the public by the Huntington's Disease Society of America and others should not be neglected as these efforts may be essential to reaching substantive change in genetic discrimination by people at risk for HD.

Implications of the findings suggest that genetic discrimination must be addressed not only from the policy aspect but also directly from the at-risk population. It is possible that personal resilience may be strengthened in those likely to encounter the more formal, as well as informal, and perceptions of experiences of genetic discrimination (legal) and societal stigmatization (interpersonal) that may ultimately lead to loss of privileges in society (Goh et al., In press).

The finding that not all reports of potential stigma or genetic discrimination were negative is novel to this study so far as the authors could determine. The actions described by participants are not novel, but are similar to the general strategies to manage perceived actual or threats of genetic discrimination (Bombard et al., 2007). In addition, this report provides new evidence of what others do that may positively influence the outcome of the event. In this study, some participants recounted perceptions of experiences where others averted or changed the process in such a way that genetic discrimination did not happen, as well as when others were believed to have increased the likelihood that discrimination would occur. Both of these instances require further examination to fully understand how these interactions could lead to positive outcomes.

The data collected in the I-RESPOND-HD study can inform a number of queries about genetic discrimination as we move forward into a legal landscape in the United States where the Genetic Information Nondiscrimination Act of 2008 (GINA) promises to alter the effects of genetic discrimination in the domains of health insurance and employment (United States Congress House Committee on Energy and Commerce, 2008). It is not clear whether GINA will end genetic discrimination in either of these contexts, and it is even possible that persons at risk for genetic illness could experience increasingly nuanced or subtle forms of stigma and discrimination. Further research will be needed to assess the impact of this law and of the shifting challenges to living with the knowledge of genetic predisposition to illnesses such as HD.

Although these reports do not provide a comprehensive picture of verified stigmatizing or discriminatory experiences, they can be useful to health care providers. It is clear that perceptions of stigma or discrimination are a part of the day to day lives of some persons at risk for HD, and that outcomes of occurrences are not always negative. Reports by people at risk for HD include feelings of surprise when, what were thought to be well intentioned efforts by others, are interpreted as ultimately being harmful. Likewise, reports also reflect surprise when efforts by others were recalled as leading to a constructive or positive resolution of a potentially negative experience. These reports can provide some guidance to health care providers when helping at risk persons to become aware of the potential for unintended use of genetic information that could lead to being treated differently. These reports also suggest that sources of stigma and discrimination may not be resolved solely by changes in laws or organization policies. Discussion of the possibilities for genetic discrimination and stigma with health care providers may help people at risk for HD to prepare for navigating interpersonal relationships within which their HD at risk status may be disclosed.

This report is from a large number of people at risk for HD. Because cognitive changes can occur many years before the onset of motor symptoms, it is unknown to what extent the potential for diminished insight, memory, or other cognitive functions could be reflected in the participants' comments. Furthermore, results are limited by lack of corroborating data from key informants in the reported occurrences, and the absence of opportunity to ask for clarification or elaboration on comments. While the narrative data do not provide all the details necessary to fully understand each person's perceived experiences, they do provide important insights into the contextual, institutional, and interpersonal factors that may influence the nature and outcome of these experiences. The majority of participants in the survey, as well as those who provided narrative comments, are from the US. Thus, the themes may not fully portray perceived experiences of people at risk for HD from Australia and from Canada. This has stimulated focus group–based research currently being undertaken in Australia. The majority of quotes provided by respondents are from females and American participants, who reflect composition of the study sample (Erwin et al., 2009).

In summary, we present in this paper evidence of the dynamic nature of social interactions and effect of perceptions of circumstances and behaviors that were both helpful and harmful in mitigating perceptions of genetic discrimination. People do not distinguish between stigma and discrimination. These phenomena are endemic to the human condition and do not respect boundaries, cultures, or gender.

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TABLE I

Demographic characteristics of at-risk participants completing the I-RESPOND-HD survey on genetic stigma and discrimination

	PREDICT	PHAROS	Total
	148	43	191
Gender N (%)			
Female	102 (68.9)	34 (79.1)	136 (71.2)
Male	46 (31.1)	9 (20.9)	55 (28.8)
Country N (%)			
Australia	52 (35.1)	0 (0.0)	52 (27.2)
Canada	12 (8.1)	9 (20.9)	21 (11.0)
United States	84 (56.8)	34 (79.1)	118 (61.8)
Education N (%)			
High School or Less	28 (18.9)	3 (7.0)	31 (16.2)
Some College or University	30 (20.3)	6 (14.0)	36 (18.8)
2-year Degree	13 (8.9)	6 (14.03)	19 (9.9)
4-year Degree	30 (20.3)	9 (20.9)	39 (20.4)
Post-grad Education or Degree	30 (20.3)	15 (34.9)	45 (23.6)
Age Mean (SD)	42.15 (10.76)	47.86 (8.37)	43.44 (10.52)

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TABLE II

I-RESPOND-HD Respondents Providing Narrative Data

	Yes	No
PHAROS	118 (95.93%)	5 (4.07%)
PREDICT	294 (94.84%)	16 (5.16%)
Subtotal	412 (95.15%)	21 (4.85%)
TOTAL	**433**	

TABLE III

Respondents for Illustrative Quotes

Paper#	Country	Study	Gender
1	USA	PREDICT	F
2	USA	PREDICT	F
3	USA	PREDICT	M
4	AUSTRALIA	PREDICT	F
5	USA	PREDICT	F
6	USA	PREDICT	F
7	USA	PREDICT	F
8	USA	PREDICT	F
9	USA	PREDICT	M
10	USA	PREDICT	F
11	USA	PREDICT	F
12	USA	PREDICT	F
13	USA	PREDICT	M
14	USA	PHAROS	F
15	AUSTRALIA	PREDICT	F
16	AUSTRALIA	PREDICT	F
17	AUSTRALIA	PREDICT	F
18	USA	PREDICT	F
19	CANADA	PREDICT	M
20	USA	PHAROS	F
21	AUSTRALIA	PREDICT	F
22	AUSTRALIA	PREDICT	F
23	USA	PHAROS	F

TABLE IV

Themes in Stigma & Genetic Discrimination Experiences

CORE THEME: Information Control
Theme: What They Encountered
Institutional Factors
Policy for Insurer
Policy for Health Care Provider
Interpersonal Factors
Prying
Stonewalling
Poor Treatment by Family
Poor Treatment by Friends
Theme: What They Felt
Sense of Injustice
Impotence
Fear
Stirring to Action
Theme: What Others Did
Advocacy
Prevent Discrimination
Perseverance to Reverse Decisions
Support
Theme: What They Did Themselves
Personal Policy for Decisions
Challenged Others
Changed the Situation
Guarded Privacy
Dissembled
Took No Action
Theme: What Happened
No Satisfactory Resolution
Satisfactory Resolution