article

What is in a cause? Exploring the relationship between genetic cause and felt stigma

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Purpose: Concern over stigma as a consequence of genetic testing has grown in response to the recent increase in genetic research and testing resulting from the Human Genome Project. However, whether a genetic or hereditary basis necessarily confers a stigma to a condition remains unexamined. **Methods:** We performed a qualitative interview study with 86 individuals with one of four conditions: deafness or hearing loss, breast cancer, sickle cell disease, and cystic fibrosis. The first two groups were divided approximately between people who ascribed their conditions to a genetic or hereditary cause and those who did not. **Results:** Respondents interpreted genetic or hereditary causes and nongenetic causes in a variety of ways. Subjects with breast cancer reported the most consistently negative interpretation of genetic cause. This response concerned future ill health, *not* an enduring sense of stigma. Deaf and hard of hearing subjects provided the most consistently positive comments about a genetic or hereditary basis to their condition, casting familial hearing loss as a vital component of group and individual identity. Respondents with sickle cell disease and cystic fibrosis offered similar and positive interpretations of the genetic cause of their condition insofar as it meant their conditions were not contagious. **Conclusions:** Although some subjects report feeling stigmatized as a result of their condition, this stigmatization is not uniformly associated with the condition's cause, genetic or otherwise. Instead, stigma emerges from a variety of sources in the context of the lived experience of a particular condition. ***Genet Med* 2006:8(1):33–42.**

Key Words: *felt stigma, genetic testing, disease cause, genetic conditions*

The increase in genetic research fostered by the Human Genome Project has heightened concerns that labeling conditions as genetic will result in increased stigmatization of these conditions and of the people who test positive for them.[1–8] There are compelling reasons for these concerns. Substantial research on the illness experience shows that the sick often find themselves subjected to negative stereotypes and isolation.[9–13] A smaller, but growing, body of work suggests that genetic conditions can trigger such responses.[2,4,14–19] But precisely why genetic conditions should raise special concerns is unclear. The historical link between genetics and eugenics might account for heightened fears,[20,21] and the frequent reliance in related commentaries on examples of serious, even fatal, conditions such as Tay-Sachs, might subtly contribute as well to the belief that genetic conditions are inherently stigmatizing.

Undoubtedly, some people with some genetic conditions are stigmatized. But little in the literature addresses why genetics in itself, as the identified cause of a condition, should exacerbate stigma or uniformly confer stigma to people diagnosed with a genetic condition. To the contrary, the literature on the social interpretation of disease suggests that beliefs about whether and how a disease confers to a person a particular meaning or status are highly variable.[22,23] Furthermore, such beliefs do not result from a single attribute of a condition (such as its cause), but from a combination of attributes, including symptoms, daily burden, severity, treatment, and the social status of the people among whom it first, most typically, or most publicly appears.[24–26] The marked shift in the meaning attributed to tuberculosis, for example, from a malady of the wealthy associated with sensitive, artistic temperaments, to the scourge of the poor associated with unsanitary living conditions and poorly ventilated, overcrowded housing, demonstrates how little disease pathogenesis itself necessarily influences social interpretation.[27]

Sexually transmitted infections (STIs) pose a counter example in which stigma is closely associated with the means of transmission. Still, within STIs there exists a hierarchy of stigma among infections, for example, syphilis is more stigmatized than hepatitis B, so that cause is not the only factor shaping stigma even for STIs. Nonetheless, the association of cause with stigma is sufficiently plausible to warrant examining whether genetics in itself, as the cause of a condition, does impart negative connotations that stigmatize the conditions it triggers.

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Stigmatization is a social process that begins with distinguishing and labeling some feature of a person such as occupation, disease, or skin color. The feature is linked to a negative stereotype that is considered "deeply discrediting" by society.[28] Identifying a person with that stereotype sets him or her apart from routine social interaction and leads to isolation and loss of status.[29] For example, in the early years of acquired immune deficiency syndrome, the majority of victims in the United States were identified as homosexuals or intravenous drug users, and the virus was described as sexually transmissible. Acquired immune deficiency syndrome became associated with the existing negative connotations carried by homosexuality, intravenous drug use, and STIs, including erroneous beliefs that people associated with such behaviors were immoral, predatory, and dangerous. Regardless of how a person contracted the disease, the diagnosis activated these stereotypes and encouraged the public to shun people with human immunodeficiency virus positive diagnoses.[30,31]

The early stages of the stigmatization process are of interest here because much concern about the stigmatization of genetic or hereditary conditions is future oriented: What *will* happen as more tests become available? To answer this we need to ask: Are genetic conditions subject to labeling and negative stereotyping in some generalizable manner? An analysis of either "felt" stigma or "enacted" stigma could help answer this question. "Felt" stigma refers to the experience of the person being ostracized or victimized as a target of stigma. Felt stigma can occur even if the targeted person cannot substantiate actual instances of discrimination resulting from stigma.[32] In contrast, "enacted" stigma refers to discriminatory or biased acts committed against the targets of stigma.[33,34]

Much previous research on stigma has examined enacted stigma by measuring responses to fictional cases acted out by research staff or presented in hypothetic vignettes.[35] These studies have been criticized for reinforcing stereotypes by failing to examine routine daily interactions and by privileging the perspective of the stigmatizer over the stigmatized. Here we report data primarily on felt stigma to address this imbalance, but also to respond to concerns expressed by people who might undergo genetic testing.

To examine whether and how a genetic cause in itself might be stigmatizing, we designed a study that compared four groups of people with four different conditions. Two conditions, breast cancer and deafness, were chosen because they are recognized as having either "genetic" (in the sense of hereditary) or "nongenetic" causes. Two other conditions, cystic fibrosis (CF) and sickle cell disease (SCD), were chosen because they typically affect different racial groups in the United States, whites and blacks, and are the most common potentially lethal conditions in each group with a single gene as the identified cause. We theorized that if genetics in itself conferred stigma to conditions, respondents with genetic breast cancer or deafness and those with SCD or CF would provide approximately similar responses to queries about the significance of their condition being genetic, that these responses would be generally negative, and that, as a group, these responses would differ from those provided by the respondents with nongenetic forms of the conditions.

We conducted 86 in-depth interviews with 21 to 23 individuals from each of the four groups. Interviews with people who had breast cancer or were deaf or hard of hearing were divided approximately in half between those who attributed their condition to genetics and those who did not. The overall goal of the interviews was to produce accounts of how living with these conditions made a difference in subjects' lives. Within these accounts, we sought to identify what role, if any, the condition's cause played in shaping experiences or in the significance attributed to the condition's perceived cause.

MATERIALS AND METHODS

Participants

We recruited study volunteers in southeastern Pennsylvania, the San Francisco Bay Area in California, and east-central Alabama, in the area of Tuskegee. We distributed flyers at numerous locations including clinics that specialized in SCD or CF and at community meetings and at schools for deaf and hard of hearing individuals. In addition, we posted recruitment announcements on websites associated with support groups for people with CF and women with breast cancer. We sought women with breast cancer who were between 1 and 5 years past their initial diagnosis, because sufficient time needed to pass for them to experience some of the issues we were examining and yet too much time might have eroded important memories. We sought to balance male and female respondents among the CF, SCD, and deaf or hard or hearing volunteers, and to keep the CF and SCD volunteers within approximately the same age range. Volunteers were interviewed in their homes, in clinics, or at a research facility. All subjects were over 18, and all were paid $25 to participate in the interview.

Interview

The same interview guide was used for all subjects, with minor adjustments made for each of the four groups. Interviews were conducted by researchers with some personal knowledge of one of the four conditions (either because she or a close relative had the condition or because she had extensive work experience with people who had the condition). An interviewer conducted interviews only in the group with which she had this expertise. For deaf or hard of hearing subjects who preferred to be interviewed in American Sign Language (ASL), the interview was translated into ASL by research staff fluent in ASL and English and familiar with social science research. Team members who had not participated in the translation reviewed it and made minor revisions. A pilot interview with the draft instrument was videotaped and reviewed, and used to create a final version of the ASL interview.

Interviews with subjects who had CF, SCD, or breast cancer were audiotaped. Some interviews conducted with deaf or hard of hearing were also audiotaped. Those conducted in ASL were videotaped and translated into English, and the translations

were audiotaped. All audiotapes were transcribed. The interviews lasted between 1 and 3 hours. This research was approved by the institutional review boards of the University of Pennsylvania and Stanford University. All subjects participated in approved informed consent procedures.

The interview guide was piloted extensively with volunteers from each of the four groups and revised several times until we determined that its questions were relevant to each group and approximately equivalent across groups. The final interview guide was a semistructured tool that included both short answer and open-ended questions. (An interview guide is available at www.geneticsinmedicine.org) The interview addressed three broad topics: (1) what the subject did to accommodate the medical and practical demands of his or her condition; (2) how the subject understood the condition's cause and whether and why this understanding had changed over time; and (3) how the condition did or did not make a difference in the subject's daily life, including family interactions (birth family and adult family), work, friendships, community activities or hobbies, and routine public interactions, such as shopping or using public transportation.

The interview guide did not use the word stigma and interviewers were cautioned against using it unless subjects spontaneously did so in their responses. This choice reflected findings during pilot interviews that some people did not know what the word meant, whereas others found questions using the term insulting because they seemed to imply that respondents should feel stigmatized. Instead we solicited responses that addressed the extent and nature of stigmatization by asking subjects to comment on the negative or positive valence of an interaction they had described during the interview, whether the interaction had triggered feelings of pride or shame, and whether it resulted in a greater sense of distance or intimacy with the people involved.

Coding

Interview transcripts were imported into *QSR NUD*IST V.6*, a qualitative data analysis software program.[36] This software was used to review and organize salient passages in the transcripts and to apply numeric codes used to generate both qualitative and quantitative reports from the interviews.[37] A two-level coding scheme was developed. First-level coding was based on answers to close-ended questions, such as whether a subject had undergone genetic testing. Second-level codes for responses to open-ended questions were developed through an iterative group process.[38] Guided by P.S., the group read selected interviews and identified the emergent themes concerning how subjects experienced living with their conditions. Both levels of coding were gathered into a preliminary codebook that supplied examples of codes and instructions for evaluating the fit between text and code and for deciding when and how to choose among multiple relevant passages.

The resulting codebook was applied to another sample of interviews and was revised. Interviews were recoded until the codebook was judged to adequately capture the interview material and a high intercoder reliability was reached.[39] Coders were trained using the interviews that had been analyzed in creating the codebook. Each interview was coded by two coders independently, who then met, reviewed coding, and addressed disagreements. If the coding pair could not agree on how to code a particular passage, the issue was referred to a weekly meeting attended by all coders and the project manager. If this group could not resolve the problem, the passage was omitted from analysis. This method, termed multilevel consensus coding, was developed by the authors, has been applied in multiple settings,[40,41] and meets accepted standards of validity in qualitative research.[42,43]

This article reports data from the section of the interview that focused on respondents' understanding of the cause of their condition. It also draws on passages coded to the more general themes of identity formation and family relationships. The choice to focus on these codes stemmed from our interest in the possibility of stigma associated specifically with the *cause* of the condition, not with the general fact of having a condition.

RESULTS

Participants

Participants are described in Table 1. The desired, even balance of male and female subjects was difficult to achieve, and final proportions ranged from 26% for males among deaf and hard of hearing subjects to 40% for respondents with SCD. Respondents in the SCD and CF groups were of approximately similar ages. For both groups more than 70% of the subjects were between 23 and 42 years old, and the average age was 36 and 34 years, respectively.

Assigning subjects to genetic and nongenetic categories

Assignment was made to "genetic" or "nongenetic" categories on the basis of a subject's understanding of his or her conditions, not on medical opinion or medical records. For the purposes of this study, we made a distinction between hereditary or genetic causes and other nongenetic or environmental causes of a condition. This distinction parallels that proposed by a recent in-depth study of lay models of disease inheritance.[44] Assessment of subject understanding of cause was based on responses to questions about what they thought caused their condition. Respondents whose answers included the following three concepts were coded as considering their condition genetic or hereditary: (1) the cause was internal to him or her (not caused by a foreign or external agent); (2) the cause was physiologic (not spiritual, mystical, or unknown); and (3) the cause was passed down to them from or through their parents. On the basis of these criteria, all respondents with CF and SCD were coded as genetic, as well as 8 of the 22 subjects with breast cancer, and 12 of the 23 subjects who were deaf or hard of hearing.

Categorizing deafness or hearing loss as genetic or nongenetic was not customary for most of these respondents. More typically, deaf or hard of hearing respondents distinguished people as members of extended or nuclear families in which deafness was common, known as "Deaf of Deaf," or as mem-

Table 1
Demographic characteristics

Characteristics	Breast cancer (n = 22)	Deaf/hard of hearing (n = 23)	Cystic fibrosis (n = 21)	Sickle cell disease (n = 22)
Gender				
Male	0	6	7	9
Female	22	17	14	13
Race/ethnicity[a]				
African-American	0	1	0	21
Hispanic	0	1	1	0
White	22	21	18	0
Mixed race	0	0	2	1
Education				
≤High school	4	3	8	11
Some college	9	6	5	5
College graduate	3	8	3	0
Graduate degree	6	6	5	5
Missing data				1
Condition's cause				
Genetic (hereditary)	8	12	21	22
Nongenetic	14	11	0	0
Age range (y), mean	37–79, 57	24–65, 40	18–47, 34	18–53, 36

[a] Race/ethnicity was assigned on the basis of subject self-description, except for those listed here as "mixed race." Their self-descriptions included German/Jamaican, German/Japanese, and Dominican/White.

bers of families in which the respondent was the sole (or almost sole) known deaf or hard of hearing person. The "Deaf of Deaf" were likely to be deaf as a result of autosomal dominant genetic mutation. However, those who were the only known deaf members of their family might have become deaf either because of a childhood illness or other environmental assault (i.e., nongenetic cause), or because of an autosomal recessive mutation. Reflecting our interest in the respondent's understanding of his or her condition, however, assignment of these subjects still relied on the three factors listed above, especially the third one concerning the subject's statements about whether his or her parents had passed down the subject's deafness or hearing loss.

Interpreting genetic versus nongenetic causes

Data contrasting the interpretation of genetic and nongenetic causes of breast cancer and of deafness or hearing loss were collected from subjects who attributed their condition to one or the other type of cause (Table 2). Analogous data contrasting the interpretations of genetic and nongenetic causes for respondents with CF or SCD were collected by asking these respondents about significance of the cause of their condition (Table 2) and by asking a hypothetic question about whether and how it might matter to them if their condition were not genetic. To emphasize the hypothetic status of these questions to respondents with CF or SCD, these results are reported separately in Table 3.

Respondents reporting genetic or hereditary causes for breast cancer provided predominantly negative interpretations of their condition and its cause. Their responses suggested that genetic breast cancer implied a broader vulnerability for themselves and for relatives, an accurate inference because cancer risk is in fact much higher for those with a predisposing mutation. Subjects expressed this heightened concern not just as it related to breast cancer itself, but also to the consequences of management strategies associated with genetic breast cancer risk, such as prophylactic surgery.

> *In the beginning I felt tainted. Not that I was less worthy, but I have cancer. And then when I found out I had the gene, it was like, a gene! I felt like I wasn't a healthy person. . . . [Having the gene is] so much more complicated than not having the gene to me because now I know I have to go through all these other things—surgeries and worries and passing on the gene. So to me it was much worse. It's like I got dealt a really bad card. (B1582)*

The only recurrent positive statement about a genetic diagnosis among these subjects was that it alleviated a feeling of personal blame for causing the cancer. As one subject commented,

> *It [a positive genetic test result] means that I didn't cause it. (B1089)*

Respondents attributing their breast cancer to nongenetic causes, especially those who had a relatively easy disease

Table 2
Interpretation of genetic and nongenetic causes (listed in order of frequency reported)

Genetic, or hereditary, cause Statements about genetic cause by people with genetic conditions	
Positive interpretation	Negative interpretation
No one is responsible (B, C, S)	Hesitant to have children (C, S)
Part of who I am (D, S, C)	Increase risk to family members (B, S)
Control/manage condition (C, S)	More medical decisions (B)
Related to beneficial mutation (C, S)	Anticipates diagnosis (B)
Better communication skills (D)	Related to harmful genetic mutations (B)
Connected to family (D)	Anger at parents (S)
Confirms continental ancestry (S)	
Could prevent with testing (S)	
Nongenetic condition/nongenetic cause Statements about nongenetic cause by people with nongenetic conditions	
Positive interpretation	Negative interpretation
No risk to relatives (B)	Limited communication with family (D)
Knows children's likely hearing status (D)	Caused own condition (B)
	Poor communication skills generally (D)
	Family does not accept condition (D)

B, breast cancer; D, deafness or hard of hearing; C, cystic fibrosis; S, sickle cell disease.

Table 3
Interpretation of hypothetic nongenetic cause: Cystic fibrosis and sickle cell disease (listed in order of frequency reported)

Statements about hypothetic nongenetic cause by people with cystic fibrosis or sickle cell disease	
Positive interpretation	Negative interpretation
More likely to have children (C, S)	Might be contagious (C,S)
Parents might feel less responsible for subject's condition (C, S)	Might be responsible for condition (C, S)
Less apprehensive about health generally (C, S)	Harder to adapt to condition mid-life than to have from birth (C,S)
	Might not know cause (C, S)

C, cystic fibrosis; S, sickle cell disease.

course, in the sense that the cancer had not recurred and they had had lumpectomies instead of mastectomies, attributed their relatively less severe disease in part to an apparent lack of a genetic or hereditary cause of their cancer. These women were more likely, however, to see the lack of a genetic cause as evidence that they had in some way contributed to their cancer, for example, by leading a stressful life.

I blame myself for the stress. (B1970)

Respondents from both groups interpreted their cancer risk as implying increased risk for female relatives. The magnitude of concern for relatives seems linked as much with severity of illness as with cause. Comments from both groups about responsibility focused on the duty to instruct relatives about cancer risk rather than on personal responsibility or guilt.

Subjects who were deaf or hard of hearing interpreted genetic and nongenetic causes differently than respondents with breast cancer. For "Deaf of Deaf" respondents, hearing loss was strongly associated with having family members who were deaf or hard of hearing, and with whom one could easily communicate and build a community, and who were supportive and insightful about how to cope with the hearing world. One subject spoke at length about her schoolmates' envy that she lived in a "Deaf of Deaf" household.

They [classmates] would want to come to my house for the weekend, and they would take turns to come to my house. . . . [O]ne time I went to somebody else's house to see why it is people wanted to come to my house, and it made it very clear. Sitting around the table with the hearing family there was absolutely no communication, just a pat on the hand and a gesture here and there. That's when I realized the kind of life I had compared to others. Around my dinner table we all sat around in a circle, in certain places [so we could sign to one another] and communication was flowing. (D3265)

Comments about hearing loss that seem to result from nongenetic causes were generally negative and focused on lack of family acceptance and limited intrafamiliar communication. One young woman described an ongoing debate she had with her mother over getting a cochlear implant, which the respondent did not want because the speech and auditory training required to adapt to it were too time consuming and would detract from what she described as her "quite full" life.

My mother just wants, has always wanted me. . . . She would work hard, she would do anything to be able to get me back my hearing. She just said that to me. She will do anything. We have argued about the cochlear implant at home. (D3436)

Respondents with CF and SCD commented on the genetic basis of their conditions in primarily positive terms that focused on responsibility and control. A genetic cause meant to them that no one was responsible for giving them the condition through contagion or neglect, which, in turn, meant that the condition did not mark them as inferior or unwanted.

It's not like my mom took some poison when she was pregnant and [gave me CF]. It's just an innocent genetic trait passed, like my eye color and my fingers or anything like that. (C2061)

They also cited the genetic cause as giving them a greater sense of control over their condition in that they knew what caused it and, having had the condition their entire lives, knew how to manage it.

R: I think part of my success with staying healthy is my mental, is the fact that it is all me. You know, it is all inside. It's like you know I still feel like I'm in control because you know it's not an outside force. . . . I don't define it as a disease because it's like I'm geneti-

cally different. So I think in that respect I would feel that it was a little more of a battle or something to fight against if it was like something from the outside. For me it's now sort of like an athletic challenge. Like a weight lifter, I can lift 300 pounds or I can shave 5 more seconds off of my mile. And for me it's sort of like an athletic challenge because it is me. I can make it to where I can breathe clearer today.

I: You are challenging yourself.

R: Right and if it was something, you know, a toxin or something I was exposed to, I think it would be more, I think it would change the tenor of it because of it would be more fighting outside forces. (C2140)

A young man with SCD commented,

I feel like it just, you know, I feel better, because I understand where it came from, I understand it. My mother had the trait, and I understand my daddy had the trait. (S4600)

Several people with SCD and a few with CF cited as a benefit the belief that the mutation that caused their condition also protected them against another disease, including malaria for SCD and cholera for CF.

A 30-year-old woman with CF mentioned its link with cholera while discussing her parents' post-World War II childhoods, her father's in Germany and her mother's in Japan.

That's like one of the things they have in common . . . born in 1940, so, they didn't really remember their early years, but stories of him in Germany were not . . . [It was] not easy to live. So, I, I obviously now understand the genetics and so on, and, in many ways, I feel like my parents are genetically very, very strong. I mean, they survived extreme malnutrition and may, I understand the science of CF is if you have the gene, you, you may have a, resistance against cholera, and that could have helped them survive. You never know, and then so... so we're, we're stuck with this, but, in many ways, genetically, it gave them, perhaps, an advantage. (C2061)

For respondents with SCD, comments about the mutation's protective function against malaria sometimes were part of more general comments about how SCD linked them to an African ancestry.

SCD is a black disease. But if you study it you know that it's a regional disease. We also know that it's based on, it's a survival disease. It is a disease that was intended by God first never to happen. But only the trait. Because the trait fought malaria . . . since we know life was created in Africa first, this had [to be] the place to be able to develop so that it became the disease to prevent His population from dying. (S4811)

Another respondent, discussing her experience of being diagnosed with SCD in 1958 and being told it was a "Negro" disease, voiced a similar sentiment about the link of SCD disease to her African ancestry, although she did not link SCD with malaria.

I was very proud. I was very proud. I mean, even though sickle cell is a painful disease and, and kind of annoying, it's a very proud heritage disease to me. I know I'm African, and I know I have a long lineage. So I was very proud. I'm still proud. (S4999)

Negative comments about a genetic cause by respondents with CF or SCD addressed deliberations over having children and disappointment that their parents had failed to prevent the disease, a sign perhaps, of ignorance on their parents' part.

Nobody knew of it. I mean, the only time genetics really came into play was when [my husband] and I were talking about having kids, and the fact that actually he has a very high probability of being a carrier. Then those questions came in: what would we do if we had a child with CF, 'cause we were going to do a surrogate, and he and I differed. We've dramatically differed on how it would all end. Where I absolutely would not want to raise a child with CF, and he felt differently about that, 'cause I think... see, he . . . he's very optimistic, and he's a little unrealistic. (C2299)

So when I couldn't get to do other things or when things would go wrong, when I did get sick and would go into crisis, I did ask that question, "why me?" Why didn't mommy and daddy know that this was going to happen? You always think that your parents know everything, that they could solve every problem. So why didn't they see this coming? Why didn't they change things, why did this come to me? Why not my brother? Why not some other family? Those things really came up a lot. (S4446)

In response to hypothetic questions to subjects with CF or SCD about what it might mean to them if their condition were not genetic, several answered that it would make no difference because the condition would be the same medically or physically, which mattered most in their daily lives. Many respondents, however, responded negatively to the hypothetic scenario, explaining that a nongenetic cause would take away important aspects of how they thought about their condition, including primarily that they were not responsible for it. These respondents also voiced concern that a nongenetic cause might deprive them of being able to say that their condition was not contagious, an important fact in interactions with new acquaintances.

[To say it is genetic] maybe it makes it a little more . . . special, is not the right word, but I can dismiss it when I always say it's genetic. It's "Don't worry about it; you're not going to catch it from me; I'm not gonna get you . . . [sick]." (C2299)

Some people when they find out that the only way you could get it is through genetics, I guess a lot of them are kind of relieved a lot of times. Because they feel like they don't have to seclude themselves from me. (S4446)

When you have a child with, even, cancer, its "Did you have an x-ray when you were pregnant?" Or, "did you smoke?" There's always ways that the public labels guilt or innocence to people, in an unfair way. And when you just tell them you have a genetic disease, you were born with it, people are so much more compassionate. (C2061)

Responses to the hypothetic scenario of nongenetic CF or SCD included two that were positive, explaining that they might feel more free to have children.

I *don't think I would be so apprehensive about having kids if I knew it wasn't a genetic problem, that it was something that you got if you were exposed to something or you contracted it through*

some ill behavior or something. If it wasn't a heredity disorder, you know I would probably have children by now. (S4646)

Reaction of friends, family, and acquaintances to genetic cause

The interviews also produced accounts of how friends, family, and acquaintances reacted to learning that the subject's condition was genetic (Table 4). These passages are useful to examine for two reasons. First, respondents sometimes used these passages to explain how they felt about people's reactions to them, an important component of feeling accepted or shunned. Second, although these are not firsthand accounts from a person encountering someone with a genetic condition, they provide rough indicators of what happens in such encounters, and thus of possible enacted, in contrast with felt, stigma.

Subjects with CF and SCD reported that acquaintances who did not know them well were relieved typically to learn that their condition was genetic because the acquaintance had been concerned that the illness might be contagious. In contrast, family members of respondents with CF, SCD, or breast cancer sometimes took the news as a wake-up call that they might be a mutation carrier and could consider genetic testing.

I've had people with and without breast cancer, they might have a distant or a close family history, say to me "You know what? I think I should get tested for the gene." It was like a light bulb went off. (B1582)

Subjects with breast cancer also reported the reverse reaction by some acquaintances. These acquaintances welcomed a genetic explanation because it allowed them to treat the subject's illness as immaterial to their own breast cancer risk. Nongenetic, sporadic breast cancers apparently could not be so easily dismissed. Less is known about what causes sporadic cancers, and a friend's nongenetic breast cancer might indicate something about the acquaintance's own risk.

[Finding out that someone's breast cancer is genetic] makes them feel happier. "Oh, I didn't have breast cancer in my family. Oh, I'm safe." . . .You know, mostly people ask [what caused my cancer] because they're concerned about themselves. I don't want to sound too negative about people, but, you know, it raises that fear. (B1369)

Table 4
Family and friends' response to genetic cause (Listed in order of frequency reported)

Family/friends'responce to GENETIC condition/cause	
Positive interpretation	Negative interpretation
Not contagious (S, C)	Parents feel guilty (S, C)
Could be a wake up call (S, C, B)	Spouses do not want children (C, S, D)
Not from neglect (S, C)	Worried about risk to other family members (B, S)
Means that I won't get it (B, S)	Attributed to irresponsible parents (S)
Intriguted, want to learn more (D, C)	Shameful (S, C)

B, breast cancer; D, deafness or hard of hearing; C, cystic fibrosis; and S, sickle cell disease.

Deaf and hard of hearing respondents who were categorized as "Deaf of Deaf," and thus likely to have a genetic cause to their deafness, reported that relatives were excited to learn that the respondent's hearing loss was genetic because it suggested that future children would also be deaf, thus adding to a network of deaf or hard of hearing family members. These respondents also reported that sometimes their hearing acquaintances were intrigued to learn about whole families who were deaf and eager to find out about growing up and living in such a setting. Respondents with CF and SCD reported a similar curiosity among acquaintances about the genetic nature of their problem.

They think that [genetic deafness] is fascinating, you know, sort of like an oddity. (D3816)

[When I tell people that SCD is genetic, they say] "Oh, really? I didn't know that." And, that they are amazed. Because, usually, usually the people that ask me are Caucasian. They are usually Caucasian that ask me what it is and how it's caused. (S4260)

Attribution of deafness to genetics elicited a small number of negative reactions among friends and family of deaf or hard of hearing subjects with nongenetic deafness. One woman, who had been deaf from birth but had no known family members with hearing loss and was unsure about the cause of her hearing loss, reported that her (hearing) relatives greeted with relief the news that genetic testing showed her not to have any of the known mutations for hearing loss. The relatives had feared the possibility of grandchildren with hearing loss.

When I told my family I didn't have the gene for deafness, they were all relieved. They were happy for me, but they said, you know, if it [a future grandchild] was deaf, that's okay, but still they were relieved. (D3526)

Respondents with SCD reported more explicit negative reaction by acquaintances who interpreted SCD as signaling a flaw in the respondent's family. One man reported this incident from his childhood.

Kids run into me at the elementary school—it was like, "Oh, your family got that disease, ooh—my family don't have that." And, kids will say the cruel—we can say the cruelest things to each other. I've had this one person [a classmate] say: "Oh, you are a lowlife, because your family has [SCD]. I thought he was crazy, because I was like, but [he said your] "parents—they got it in your family—I don't have that and I can never catch that." (S4407)

A respondent with CF reported a negative response by her grandmother because the respondent's CF diagnosis challenged the grandmother's account of the family's ancestry. The respondent is of German-Japanese descent, and the grandmother in the story is Japanese.

[W]e have the delta 508 [mutation] and then for many years an unidentified one, and the delta 508's probably from my dad; he's German and then finally they found another Japanese mutation.

But it ended up being like very common. So then we started to tell my grandmother, maybe you have Russian blood. And she was so offended, because Japanese and Russians, you know, have their wars, and she didn't want to think that her blood was mixed with [the Russians].

[A doctor] drew our blood, and he drew my grandmother's blood. She did not have the gene, and my grandfather was killed in the war. He died in 1940. So grandma's like, "Oh, I'm innocent. I'm purebred Japanese." She comes from like a Samurai family, so it's like very . . . stigma, stigma. . . . (C2061)

DISCUSSION

The concern that increased genetic testing will result in stigmatization of the conditions tested for and of people whose results are positive depends on the assumption that a genetic cause confers a uniform meaning to conditions. We investigated this assumption from the perspective of felt stigma, which refers to the experience of the person being ostracized or victimized as a target of stigma. Our research has shown that a genetic cause did not automatically impart to respondents a sense of being stigmatized. We hasten to emphasize that this does not mean that respondents might not have reported feeling marginalized as a result of having the conditions that we examined. Rather, it means that that they did not associate such feelings with the condition's cause.

Each group of respondents provided both positive and negative interpretations of genetic and nongenetic causes; however, each also tended to speak more favorably of one type of cause. All but the breast cancer group cast the genetic cause more favorably. The two groups who differed the most from each other and from the other study subgroups were the respondents with breast cancer and those who were deaf or hard of hearing. Breast cancer respondents provided the most consistently negative interpretation of a genetic or hereditary cause both in its implications for their own health and for its disturbing associations with ill health more broadly for themselves and for relatives. These respondents, however, reported only one example that constituted felt stigma associated with cause. One woman stated that having been diagnosed with genetic breast cancer made her feel "tainted." This feeling of being tainted occurred at the time of diagnosis and was not described as enduring much beyond diagnosis. Overwhelmingly the most consistent source of feelings of shame or fear of rejection for respondents with breast cancer came from the bodily disfigurement they had as a result of breast cancer treatments. (Respondents did not associate these comments to cause and they are not described here.)

In contrast with findings reported by other studies that suggest a high degree of guilt associated with genetic breast cancer over the possibility of passing down the mutation to their children,[45–47] respondents in this study who believed their cancer to be genetic did not emphasize such feelings. Nonetheless, they did express a sense of urgency that their female relatives exercise considerable vigilance in monitoring their risk for developing breast cancer and took as a serious personal responsibility the need to inform relatives about potential risk.

Deaf and hard of hearing subjects provided the most consistently positive interpretation of genetic or hereditary cause because for many it stood for growing up among people with whom they could communicate. Responses from these subjects demonstrated how individuals can incorporate a genetic condition as a positive feature of identity. As "Deaf of Deaf," these respondents regarded their deafness as normal and valued.

It is possible that these positive responses came primarily from individuals with autosomal dominant hearing loss, which would account for deafness being common among family members. Individuals with autosomal recessive hearing loss would be unlikely to have multiple deaf family members, especially in their extended families, and would likely experience the same kind of limited communication as individuals with deafness caused by illness or other environmental assault. Thus, the positive feelings associated with respondents in this study categorized as having hereditary hearing loss might more precisely be associated with autosomal dominant hearing loss rather than with genetic hearing loss generally.

The difference between respondents with breast cancer and respondents who were deaf or hard of hearing is logical. Although the first is a serious, potentially fatal disease, the second is a condition that is contextually a disability (when dealing with the hearing world) and one that can be accommodated, albeit not always easily or successfully.

Respondents with SCD and CF reported approximately similar interpretations of the genetic cause of their condition, with the exception that only among the respondents with SCD were there reports of negative attributions by others on the basis of a genetic cause. These reports were limited to two people, and in both cases to incidents from the subject's childhood. Because we tried to present examples that were typical of findings from each of the four groups, one related finding might have been obscured. People with CF were not only less likely to report negative experiences or feelings about having a genetic condition, but were also more likely to cast the genetic cause in clearly positive terms. This positive framing was less pronounced than among the deaf or hard of hearing, but still greater than those with SCD. Further analysis will examine this difference as possibly being associated with racial identity. Respondents with SCD often associated their condition with being African-American, and then sometimes with discrimination, whereas those with CF rarely mentioned any link between being white or of European descent and having CF.

Another element in the SCD and CF responses about causality is important to note. When asked to discuss genetic versus nongenetic cause, these respondents emphasized the benefit of having had a *genetic* condition from birth and being allowed to adapt to it. They contrasted this negatively with a hypothetic nongenetic cause, typically an infection, which implied that negligence had somehow contributed to their condition. However, the differences they highlighted might not have been so much between genetic and nongenetic as between

congenital and noncongenital, a difference that further research could examine.

This study suggests that people interpret a genetic cause (presumably any cause) in the context of their lived experience with a particular condition. What people bring to that interpretation extends beyond a technical description, such as *transmission through reproduction*, and incorporates aspects of daily life associated with their conditions. Thus, "genetic cause" did not communicate to respondents in this study a univocal message about defect, responsibility, or innocence, as some have supposed.[6,7] Rather, people interpreted "genetic cause" variously as the basis for family cohesiveness (deaf), as a link to one's ancestry (SCD), or as a sign of being tainted (breast cancer).

There are several limits to this study. First, our respondents were volunteers and thus might be assumed to have come to terms with the conditions they have in a way that those who did not volunteer might not have. This selection bias might account for the generally affirmative stance toward the challenges of living with these conditions reported by respondents. However, there is no obvious reason that those who have not come to terms with a condition would offer interpretations that would contradict our primary finding that people do not uniformly interpret the significance of a genetic cause. Still, those who did not volunteer if given the opportunity might express a proportionately greater number of negative interpretations. This limit could be addressed in a larger study that solicited a broader range of participants. An important distinction needs to be highlighted. This study did not find that respondents never, or only infrequently, experienced felt stigma associated with their conditions. Rather, the finding is that *felt* stigma typically was not related to the *cause* of their conditions. In other sections of the interview, for example, in sections discussing the demands of medical treatment, respondents often expressed complaints and resentment, and described difficult, alienating experiences associated with their conditions.

A second limitation is that we examined only four conditions. Because our results suggest a good deal of variation across conditions, one should be cautious in extending these results to other conditions. This caution against treating these findings as generally applicable to all genetic conditions also can be seen as instructive. It emphasizes that the significance of a genetic cause, at least for the people who have such conditions, is not fixed but is strongly influenced by social consequences associated with the cause. These consequences are actively created by language, attitude, and practices, such as those that deaf or hard of hearing subjects recount about their families, or such as those that might occur between practitioners and patients.

The long history of the misuse of genetics to justify social discrimination suggests caution in dealing with genetic information. But data from our study suggest it would be a mistake to *assume* that stigma is always and automatically associated with genetics and that perhaps even doing so contributes to the negative attitudes associated with these conditions.

Because predisposition and presymptomatic testing is rapidly outpacing testing for single gene disorders such as CF or SCD, it is important to consider what implications this research might have for that testing. There is now no popularly recognized independent category for people who have gone through predisposition and presymptomatic testing. As more people do have such tests, categories will emerge and they will have associated with them a range of attributes and meanings. These are likely to vary considerably by condition, and between insiders and onlookers, and thus to be expressed differently in felt versus enacted stigma. In the meantime, to the extent that interpretations of genetic cause are actively created and not inherent, what these findings suggest is that practitioners and the medical establishment hold a powerful position. The more precise practitioners can be about the significance of genetic cause, and the more careful policy makers can be to not automatically associate stigma with genetic testing, the more they will be able to contribute to restricting the assumption that genetic testing must be associated with stigma and thus to open the way to novel, less negative interpretations of this valuable medical technology.

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