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## What's at Stake? Genetic Information from the Perspective of People with Epilepsy and their Family Members

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### Abstract

Substantial progress has been made in identifying genes that raise risk for epilepsy, and genetic testing for some of these genes is increasingly being used in clinical practice. However, almost no empirical data are available from the perspective of people with epilepsy and their family members about the impact of genetic information and potential benefits and harms of genetic testing. To address this gap we conducted in-depth qualitative interviews with 40 individuals (22 with epilepsy, 18 unaffected) in the USA from families containing multiple affected individuals who had participated in epilepsy genetics research. The interviews were coded and analyzed using the principles of grounded theory. Several major themes emerged from these interviews. Participants expressed “personal theories of inheritance” that emphasized commonalities among relatives and the idea that disease risk is most shared by family members who share physical or personality traits. Most participants said they would have genetic testing if it were offered. They cited many potential benefits, including learning what caused epilepsy in their family, being better able to care and advocate for children at risk, reducing guilt and blame, providing an increased sense of control, and relieving anxiety in unaffected individuals who test negative. The influence of genetic information on reproduction was a particularly salient theme. Although respondents believed genetic testing would be useful for informing their reproductive choices, they also expressed fear that it could lead to external pressures to modify these choices. Other concerns about the potential negative impact of genetic information included increased blame and guilt, increased stigma and discrimination in employment and insurance, self-imposed limitations on life goals, and alterations in fundamental conceptions of “what epilepsy is.” Consideration of the perspectives of people with epilepsy and their family members is critical to understanding the implications of contemporary epilepsy genetic research and testing.

### Keywords

genetics; stigma; epilepsy; reproductive decision making; USA; family

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Epilepsy is one of the most common neurological disorders, affecting approximately three percent of individuals at some time in their lives (Hesdorffer et al., 2011). Epilepsy is broadly defined by recurrent *unprovoked* seizures, i.e., a lifetime history of two or more seizures occurring in the absence of an acute structural or metabolic insult to the central nervous system (Hauser et al., 1991). However, it is so clinically heterogeneous that it is considered a collection of different disorders (“epilepsy syndromes”) with distinct causes (Berg et al., 2010). Seizure manifestations vary from brief lapses in consciousness to whole body convulsions, and associated sensory, motor, psychic, and other symptoms are also extremely variable (Choi et al., 2006).

Genetics has emerged as a central focus of epilepsy research, and more than 20 genes with a major influence on risk for human epilepsy have been identified (Ottman et al., 2010). The genes identified so far affect risk in a very small proportion of people with epilepsy – primarily those from rare families consistent with monogenic (Mendelian) inheritance patterns such as autosomal dominant, autosomal recessive, etc. Most people with epilepsy have no affected relatives, and most forms of epilepsy are inconsistent with Mendelian modes of inheritance. Major research efforts are also underway to identify the genes involved in these “genetically complex” epilepsies, where the relevant genes probably have only modest effects and act in concert to raise risk, possibly in combination with environmental factors (Ottman, 2005). Clinical genetic testing for epilepsy currently is available for several epilepsy syndromes in which genes influencing risk have been identified (e.g., testing for mutations in the *SCN1A* gene in Dravet syndrome, where >70% of affected individuals have mutations), and is a major focus of current consideration in the clinical management of epilepsy (Ottman et al., 2010; Pal et al., 2010).

Despite the increasing availability of genetic tests for epilepsy, little is known about how people with epilepsy understand and experience genetic information or how they perceive the potential benefits and risks of genetic testing. We argued previously that analysis of the ethical, social and political concerns raised by genetic research on epilepsy and the advent of genetic testing is urgently needed (Shostak & Ottman, 2006). Many of these concerns are not unique to epilepsy. For example, among the ethical and legal concerns raised by genetic tests in general are appropriate informed consent, confidentiality and privacy of genetic information, the imperative of balancing individual, parental, and societal interests when considering genetic testing for a minor, and the disclosure of genetic information to family members (d’Agincourt-Canning, 2001; Hallowell et al., 2003; Lapham et al., 1996). Likewise, genetic testing more broadly raises social and political concerns such as equity in the availability and affordability of prophylaxis and/or treatment for identified genetic susceptibilities (Burke et al., 2001). However, as a physiological condition and a social experience, epilepsy – like all diseases (Timmermans & Haas, 2008) – has unique characteristics that must be taken into account (Shostak & Ottman, 2006).

Of particular concern are the inter-related issues of stigma and discrimination, which social scientific studies spanning nearly three decades have highlighted as a significant part of the experience of having epilepsy (Gehlert et al., 2000; Jacoby, 2002; Jacoby et al., 2004; Morrell, 2002; Scambler, 1989; Schneider & Conrad, 1983). Some evidence indicates that individuals’ experiences of stigma have declined since WWII, at least in Britain and the United States; however survey research in those countries suggests that people with epilepsy still are often viewed as “violent, likely to go berserk, retarded, sluggish or slow, antisocial, and physically unattractive” and they are more likely to experience social rejection than persons with cerebral palsy or mental illness (Jacoby, 2002). Even in the absence of actual experiences of stigma and discrimination, people with epilepsy describe their fears of experiencing stigma and discrimination as a significant component of living with epilepsy (Scambler, 1994). Together, the physiological and social dimensions of living with epilepsy

can be quite consequential. As compared to their peers who do not have epilepsy, people with epilepsy report reduced social interactions, rates of marriage, reproductive rates, self-reported health-related quality of life, and increased psychological distress (Jacoby et al., 2004; Krauss et al., 2000; Morrell, 2002). Reducing stigma and discrimination has been a major focus of advocacy by and for people with epilepsy.

Drawing on analysis of in-depth interviews, this paper examines understandings of genetics and orientations towards genetic testing among people with epilepsy and their family members. Our goal is not to evaluate the extent to which respondents' beliefs are concordant with scientists' or clinicians' understandings of genetics or genetic testing. Rather, we consider people with epilepsy and their family members as experts capable of analyzing the burdens and benefits of genetic information and genetic testing and thereby "casting a rather different light on the contests for meaning and rationality..." (Rapp, 1998). We find that the hopes and fears of people with epilepsy and their family members with regard to genetic information reveal important aspects of the experience of living with a stigmatized illness. These include not only concerns about potential discrimination in employment and insurance, but also broader issues about life trajectories, reproductive decision-making, and how epilepsy itself is understood.

## Background and Research Questions

### The Social Consequences of Genetic Information

The emergence of the Human Genome Project (HGP) heightened longstanding sociological concerns about the social consequences of biological information (Duster, 2006). The concept of "geneticization" served as a conceptual jumping off point for many early social scientific writings about the implications of genetics. As introduced by Lippman (1991:19), geneticization refers to "an ongoing process by which differences between individuals are reduced to their DNA codes, with most disorders, behaviours, and physiological variations defined, at least in part, as genetic in origin." Lippman saw genetics as a "dominant discourse" with numerous potential negative social implications (Lippman, 1991).

There is clear evidence that people attribute a wide variety of individual health and social outcomes to genetics (Shostak et al., 2009). However, genetic information does not straightforwardly lead to genetic determinism (Freese & Shostak, 2009). Studies of different kinds of genetic tests indicate that users of genetic testing appreciate the nuances of probabilistic risk and predictive uncertainty, and are correspondingly circumspect in their interpretations of genetic information (Franklin & Roberts, 2006; Hallowell et al., 2004; Markens et al., 1999; Whitmarsh et al., 2007). People in families with a high prevalence of mental illness emphasize the interactions of genetic and environmental factors in their accounts of the etiology of disease (Meiser et al., 2005). In one of the only available analyses of how people make sense of genetic information about complex diseases (i.e., rather than autosomal dominant conditions), Lock and colleagues (2006) find that even when individuals embrace the idea that Alzheimer's disease "runs in the family," they interpret information about genetic susceptibility in the context of their own beliefs about the multiple causes of illness and observable risk factors in their families. One year after receiving "personalized risk assessments" for late onset Alzheimer's Disease, participants had "transformed" the estimates into accounts that "fit" with their experience of being related to someone with Alzheimer's disease, personal assessments of their family history, and knowledge about the disease gathered from a variety of sources (Lock et al., 2006). Nonetheless, evidence suggests that genetic information can reshape concepts about health and illness and images of people who are ill and their relatives.

## Genetics and Stigma

To date, research on genetics and stigma has focused especially on mental illness. While advocates for the mentally ill hoped that attributing mental illness to genetic causes would reduce the stigma surrounding mental illness, others argued that by casting it in more essential terms, genetic explanations would make mental illness seem less treatable and more threatening (Bennett et al., 2008; Phelan, 2005). Genetic explanations of mental illness raise the possibility that the biological relatives of mentally ill persons will experience “associative” (Mehta & Farina, 1988) or “courtesy” stigma (Goffman, 1963). Epilepsy is not a mental illness. However, like mental illness, epilepsy has long been stigmatized, and empirical research provides reason for concern about the consequences of genetic attributions for stigmatized conditions (Bennett et al., 2008; Phelan, 2005).

On the whole, research on genetic attributions and stigma suggest that genetic understandings interact with, rather than overturn, longstanding preconceptions about illnesses (Schnittker, 2008). This finding is supported by research that explores the relationship between beliefs about genetic cause and felt stigma, which demonstrates that people interpret the meaning of genetic causes in the context of their experience with the condition (Sankar et al., 2006). Moreover, it is congruent with research on the social meaning of disease more broadly, which suggests that “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” (Sankar et al., 2006: 33).

Research on genetics and stigma tends to use standard measures of stigma, such as desire for social distance, endorsement of reproductive restrictions, perceived dangerousness, and social acceptance (Bennett et al., 2008; Meiser et al., 2005; Phelan, 2005). This research theorizes stigma as a social process with multiple components (Link & Phelan, 2001; Yang et al., 2007). A different approach is to conceptualize stigma vis-à-vis “moral experience”. In this usage, moral experience refers “to that register of everyday life and practical engagement that defines what matters most for ordinary men and women” (Yang et al., 2007: 1528). From this perspective, stigma is a “fundamentally moral issue in which stigmatized conditions threaten what really matters for sufferers” in their daily lives (Yang et al., 2007: 1528). We use this analytic approach as a means of exploring a wider range “stakes” than would be encompassed by standard measures of genetics and stigma. Further, we contend that understanding stigma as a moral experience is a warrant for qualitative research methods, such as in-depth interviews (Kleinman et al., 1995) and ethnography (Yang & Kleinman, 2008), which allow the researcher better access to individuals’ local moral worlds. This perspective highlights also the importance of multiple vantage points in research on stigma, such that analysis is strengthened when it includes, for example, not only the person with illness, but also family members (Yang et al., 2007).

## Research Questions

In this paper, we address the following three questions:

1. How do people with epilepsy and their family members understand genetics and heritability?
2. How do people with epilepsy and their family members perceive the risks and benefits of genetic testing for epilepsy?
3. What do people’s hopes and fears regarding genetic testing reveal about the local moral worlds of people with epilepsy and their family members?

## Data and Methods

From 2005–2006, we conducted in-depth, qualitative interviews with people with epilepsy and their family members (n=40). The sample for these interviews was drawn from a database of families who had previously participated in Epilepsy Family Study of Columbia University (EFSCU). EFSCU is a long-term investigation of the genetic influences on epilepsy that began in the mid-1980's as a study of familial aggregation and evolved into a genetic linkage study. The primary goal of the linkage study was to identify chromosomal regions likely to harbor genes that influence risk. Families were eligible for inclusion if they contained either a sibling pair or three or more individuals with epilepsy of unknown cause, and were identified through physician referral or self-referral in response to advertisement through voluntary organizations and a study web site. A total of 97 families were included. In these families, 1070 individuals participated in research activities including telephone interviews about their personal and family medical history, review of their medical records, and donation of a blood sample. These activities were carried out between 1992 and 2007. Our protocols for the protection of human subjects in research have been approved by the institutional review boards of both the Columbia University Medical Center and Brandeis University. In the analysis that follows, interviews are identified by number to protect the confidentiality of respondents; the names that appear in quotations are pseudonyms.

Due to the EFSCU requirements, all of the people in the database come from families in which two or more individuals have epilepsy. Because the extant literature suggests that prevalence of a condition among family members can shape beliefs about genetics, we stratified the sample into two groups - one in which two people in a family have epilepsy and the other in which  $\geq 4$  people in a family are affected. Whenever possible, within each family we interviewed a person with epilepsy and an unaffected family member. More broadly, our sampling strategy was purposive, with a deliberate effort to encompass a wide range of variation in life situations, illness experience, sociodemographic characteristics (e.g., gender, ethnicity, age). In an exploratory, qualitative study such as ours, these characteristics cannot serve as a basis of formal comparison, although they provide a helpful overview of into the composition of the sample (See Table 1).

Topics explored in the interviews with people with epilepsy and their family members included the following: 1) the subject's experience of learning that he or she (and/or a family member) has epilepsy; 2) how having epilepsy (and/or having a family member with epilepsy) has affected his or her life; 3) beliefs about heritability; 4) perspectives on genetic testing; 5) social networks. Because participants live across the United States, almost all of the interviews were conducted over the telephone; they were transcribed by a professional service and uploaded into Atlas.ti for analysis.

The interviews were coded and analyzed using the general principles of grounded theory (Charmaz, 2006; Clarke, 2005; Strauss & Corbin, 1998). In this approach, the analyst codes the data by giving temporary labels to specific words and phrases from the interviews, and then compares codes across the interviews, elaborating the properties of codes and relationships between them. Related codes are "densified" into analytic categories (Clarke, 2005). Codes and categories are strengthened (and sometimes discarded) as they are compared to and evaluated against new codes and categories that emerge as analysis progresses. The codes and categories that are robust enough to withstand this iterative process are integrated to theorize the conditions and the contexts of the phenomena of interest.



## Results

We asked study participants directly about their *beliefs about genetics* and their *perceptions of the potential benefits and harms of genetic testing*. In their answers to these questions, which we describe below, two additional themes emerged. First, for people in families affected by a potentially heritable illness, genetic information may be inextricably bound up with *decisions about reproduction*. Second, and related, for people living with epilepsy, part of what is at stake in the increasing availability of genetic information is *the ontology of the disease itself*, that is, what epilepsy is and how one may live with it.

### Beliefs about Genetics

There were two direct questions in the interview that inquired about beliefs about epilepsy, genetics, and heredity. First, respondents were asked whether they think that “epilepsy is something that can be handed down from parents to children.” Second, they were asked whether they think that epilepsy is something that “runs in your family.” In addition to their responses to these questions, respondents mentioned genetics and heredity in their responses to broader questions about their beliefs about the causes of epilepsy and their experiences of living with epilepsy or being the family member of someone with epilepsy.

A majority of the respondents report believing that epilepsy is genetic and inherited. As this respondent commented, “It’s in my family. It’s hereditary. My father had it, his father had it, and it’s in my family...” (Interview 17). Related to this, respondents report believing that having a family member with epilepsy increases the likelihood of having epilepsy. The belief that epilepsy runs in the family may exist even when people with epilepsy have been told by their doctors that there is no genetic component to the condition, as in the case of this respondent who commented “I grew up with the doctor telling me that it isn’t genetic, but I believe that it is, that it is something inherent that got triggered somehow” (Interview 34). Both people with epilepsy and their family members report worrying about whether their children will have seizures. People with epilepsy report being “devastated” and “guilty” when their children were diagnosed with epilepsy (Interview 38). However, some respondents commented also that having had epilepsy makes them especially aware of and sensitive to the needs of a child with epilepsy. Only a few unaffected family members reported that they worry that they may have seizures in the future.

People in families affected with epilepsy often have detailed knowledge about each other’s seizure experiences (e.g., age of onset, types of auras preceding seizures) and they use this knowledge in formulating their perceptions of risk for themselves and their children. This dynamic is especially vivid in families where multiple family members began to have seizures at approximately the same age. For example, one respondent reported that when his son approached the age of 10 – the age at which the respondent, his brother, and his father all began having seizures – he began to watch his son “vigilantly” for any sign of seizure activity (Interview 13). People also use their observations of their family members to predict whether and when their seizures may lessen or cease, as reflected in this comment “My father’s seizures stopped when he was 30, so I figured maybe mine would stop when I was 30...” (Interview 17). Similarly, respondents watch their families for clues about how the condition may have been inherited, as this respondent reported:

My parents divorced and my dad got remarried. So we’re kind of curious, you know, if [our half sibling] end[s] up with seizures. You know, we hope she doesn’t...also-if she doesn’t, does that mean like it was what we got from my mother, because [we] don’t share the same mother with our little sister. So, you know, if she has seizures, then [we’d] probably be pretty sure that it was something we got from my dad (Interview 21).

The respondents in the 2005–2006 interviews are clearly not representative of all people with epilepsy; they are unusual in two important respects. First, they come from families in which multiple individuals are affected, whereas most people with epilepsy have no affected relatives. Second, they have made the decision to participate in genetic research, which may reflect an unusual interest in, and possibly understanding of, the genetic influences on epilepsy. However, we note that even in this sample, arguably inclusive of people most well informed about contemporary epilepsy genetics research, respondents have their own understandings of what exactly “heredity” or “genetics” means. That is, in contrast to formal scientific understandings of genetics and heredity, subjects hold “personal theories of inheritance” (McAllister 2003), which emphasize their perceptions of commonalities among family members, what Lock and colleagues (2006) call theories of “blended inheritance”. For example, when asked about her thoughts regarding the possibility that her children might develop epilepsy, one respondent said that her older son is just like her husband (who does not have epilepsy), “blond haired, blue eyed, and sweet” while the younger boy is just like her, “red haired and energetic...just like a Kennedy...he has our genes” (Interview 31). Therefore, because it is her family that is affected by epilepsy, it is the younger, redheaded son about whom she is worried. Similarly, speaking of her grandchildren, Victoria – whose daughter Anne has epilepsy and is the mother of two sons --commented that one of her grandsons “looks just like a Dixon, just like his grandpa Jack.” Jack, Victoria’s husband, has epilepsy and she believes that Jack “gave it” to their daughter, Anne. Because Jack and Anne “look like Dixons” and both have epilepsy, Victoria suspects that the grandson who “looks like a Dixon” is more at risk (Interview 15).

In summary, genetics is increasingly a part of the experience and self-understandings of many people with epilepsy and their family members. However, genetics does not dominate people’s understandings of epilepsy, even for individuals who believe that epilepsy runs in their family. Rather, genetic information is imbricated with people’s beliefs and ideas about their lives, their families, and their hopes for the future.

### Perceptions of Genetic Testing

Respondents were asked directly about their perceptions of genetic testing. The stem for this set of questions was “As you may know, scientists have identified genes which help to cause some forms of epilepsy in some people” People with epilepsy then were asked “If a blood test was available that could tell you if you have one of these genes, would you want to take that test?” The family members of people with epilepsy were asked both “If a blood test was available that could tell your family member that he or she has one of these genes, would you want him/her to take that test?” and “If a blood test was available that could tell you if you have one of these genes, would you want to take that test?” Follow up questions probed for the reasons for the answers given to these questions. Both sets of respondents were asked how genetic testing could be helpful and/or harmful to people with epilepsy or their family members. Research indicates that reported hypothetical interest in genetic testing only modestly predicts the choices individuals eventually make (Sanderson et al., 2010). However, respondents’ assessments of the possible risks, benefits, and implications of genetic testing reveal key aspects of what is at stake in genetic testing from their perspectives.

Only three people with epilepsy said that they certainly would not participate in genetic testing were it offered to them. Of these three, only one person with epilepsy indicated that she would not want to take a genetic test under any circumstances. The two other respondents with epilepsy stated specifically that they would undergo testing only as part of scientific research but would not do so for their own “personal knowledge.” As one person with epilepsy put it, genetic information was not personally compelling to him “Because it’s something that I’m not going to change. It was something I was born with and I can’t

change it” (Interview 12). The family members of people with epilepsy also tended to have a positive orientation to genetic testing, with only three respondents stating that they would not want to be tested.

Some participants indicated that they believed that as participants in the EFSCU, they already had undergone genetic testing. Although molecular genetic analysis was carried out in the study, the EFSCU consent form clearly stated that it was for research purposes only, that the study did not involve clinical genetic testing, and that participation would not allow participants to learn whether or not they carried a gene that raised risk. Such comments point to the complexity of distinguishing between molecular analyses conducted as part of scientific research and clinical genetic testing (Timmermans and Buchbinder 2010).

**Perceived Potential Benefits**—Respondents identified several ways that genetic information would be beneficial to themselves and their family members. These included knowing why they have epilepsy, being able to better care and advocate for children at risk for developing epilepsy, being able to better anticipate seizure onset. Additionally, unaffected family members reported that a negative test would reduce their anxiety. Study participants also expressed enthusiasm for the potential of genetic information to improve the lives of people with epilepsy more broadly, by leading to new options for treatment or a “cure” and raising public awareness and understanding of the condition.

Generally, people with epilepsy expressed belief in the power of knowledge itself as “the most powerful tool” for facing epilepsy, especially insofar as it increases people’s sense of control over their own lives and ability to care for their children (Interview 38). Respondents noted the possibility that genetic testing might help to identify the causes of epilepsy, in themselves and their family, thereby reducing uncertainty and self-blame and answering questions such as: “Why did I get it and not Bill Smith down the street or something?” (Interview 1). Another respondent commented that he would get some “ease” from knowing that “I didn’t do anything to get it” and having an “answer” to the puzzle of “our family history” (Interview 17).

Even absent of methods to reduce the risk of seizures, participants in this study suggested that genetic information might help parents – and doctors – quickly and correctly identify seizure disorders in at risk children. Likewise, respondents believe that having genetic information would enhance the ability of parents to advocate for their children in health care settings:

I think you might pay more attention and be quicker. If your kid started to have a seizure, you’d have a lot more information immediately. You know, you would skip the preliminaries and go right to an EEG.... If I knew that I was a carrier and my child had a situation that looked to me that it might be related to epilepsy, having a brain event, I’d be much more aggressive and much faster at the hospital to pursue care in a very particular way (Interview 34).

For people at risk for types of epilepsy that are characterized by an “aura” preceding convulsion, genetic information might improve their ability to watch for signs of seizure onset and thereby take steps to ensure their own safety before seizing:

For those who were positive they could then become better informed so that with this form of epilepsy that tends to have some sort of auditory aura that our family is experiencing they could be more aware of those advanced signs and be prepared to take action to be in a safe position or safe place. If they are driving, stop the car, pull the shoulder or something for their own personal safety. If they have any



anticipation that they might be at risk and experiencing the signs that go along with this unique form of epilepsy (Interview 39).

Lastly, people who perceive themselves to be at risk, by virtue of their family history, might be relieved of decades of concern:

Well, for those who would test negative, it would give them a sense of relief and assurance that they weren't at risk for experiencing this form of epilepsy (Interview 29).

The possibility that genetic testing would lead to better treatments, and possibly even a "cure" (Interview 22) for epilepsy, was identified repeatedly by participants as a potential benefit of genetic testing. Respondents believe generally that "the first step to finding a cure is to identify a cause" (Interview 15) and that genetic research is a "stepping stone towards treatment" (Interview 20). Indeed, even respondents who noted that they did not see genetic testing as relevant to their own lives expressed a willingness to be tested if this would contribute to efforts to help other people, and especially "future generations" with epilepsy:

It may not be for myself or my family, but it might help somebody else in the future down the road, you know, in my family. (Interview 9)

To be sure, this group of respondents is highly selected by virtue of their prior participation in biomedical research. However, other research also indicates that the desire to contribute to scientific research and medical practice is a widely shared motivation for genetic testing, especially among people in families with a high prevalence of a particular illness (Bernhardt et al., 1997; Geller et al., 1999; Henneman et al., 2004; Lewis et al., 2009; Peters et al., 2004; Phillips et al., 2000).

**Perceived Potential Harms**—Respondents also identified a number of ways in which genetic testing could harm people with epilepsy. One set of concerns centered on the potential of genetic testing to be used to stigmatize and/or discriminate against people with epilepsy and their children: "genetic testing can be abused like any private medical information can be abused" (Interview 28). Respondents were especially concerned that genetic information might be used by insurance companies to categorize epilepsy as a "pre-existing condition" even for people who have not yet had a seizure (Interview 30). They also noted the possibility of abuse of genetic information by employers making hiring decisions (Interview 36) and health and life insurance companies making policy decisions (Interview 29). Both people with epilepsy and their family members stated that they would not undergo genetic testing unless they were convinced that it would not threaten health insurance eligibility or coverage for themselves or their children. Respondents also reported that it would be critical that the test be safe and affordable.

Several respondents also expressed fears that genetic testing would lead to attempts to "eliminate people" who have epilepsy "in utero," or to discourage people with epilepsy from having children. The history of eugenics figured prominently in how people expressed these concerns. One respondent commented that genetic testing, and especially prenatal genetic testing, "conjures in my mind this whole deal about the Hitler era and the Aryan race" (Interview 13). In another reference to the atrocities of the Nazi era, a respondent stated, "I'm not going to, you know, jump into an oven, so I can purify the race" (Interview 10). Another respondent expressed concern about "researchers" taking the information "out of context" and "playing God" with it (Interview 6). As we discuss in detail below, respondents also expressed concern about how genetic information would affect how people with epilepsy make decisions about reproduction.

A third broad set of concerns about potential harms centered on whether genetic information might lead people with epilepsy, or with a genetic predisposition for epilepsy, either to “feel damned or condemned” (Interview 32) or to “limit themselves”(Interview 27), “feel pitiable” (Interview 23), “make them afraid to live” (Interview 7), or “use the genes as an excuse” not to reach their potential (Interview 19). One respondent raised the possibility that given the stigma associated with epilepsy, someone might be so upset to learn that he carries a gene for epilepsy that he might decide to “end it all, 29<sup>th</sup> floor...” (Interview 13). Respondents thought that people at risk might be “better off not knowing,” especially if knowing that they are at risk makes them “afraid of doing something” in their lives (Interview 26). A parent of two children with epilepsy commented that “if you’re sitting around and waiting for epilepsy to come knocking then...maybe you would not live your life quite the right way” (Interview 31). Respondents also expressed a somewhat different concern that genetic information might be used by people with epilepsy to justify behavior, like alcohol consumption, that may trigger a seizure:

What good is it for a person like that who has no control over her own life just to be able to say, well, it’s not my fault, it’s my mother’s fault (Interview 11).

These comments highlight the salience of concepts such as “responsibility” and “control” in the lives of people with epilepsy. Participants in this study often provided us with detailed accounts of the medications that they take, behaviors that they prioritize (e.g., getting enough rest), and “triggers” that they avoid (e.g., alcohol, exhaustion) in an effort to control their seizures. Research suggests also that parents may limit the activities of children with epilepsy, in order to avoid possible harms to a child at risk of seizure (Williams et al., 2003). These comments thus highlight a set of concerns that are likely to be of particular import to people affected by epilepsy.

Another set of potential harms concerns the experience of parents with epilepsy who may have transmitted to their children a genetic susceptibility to epilepsy. People with epilepsy reported that they expected that they would experience significant guilt if genetic testing confirmed that they had passed on to their children a genetic susceptibility to epilepsy:

I would probably feel really guilty. Even though it wasn’t my fault and all that. I would feel guilty for you know, possibly putting that... health issue at their feet. Not that I probably would have stopped having children (Interview 6).

Another reported concern was that the parents of children with epilepsy would be “blamed” by others for giving it to their children (Interview 21); however, at the same time, a participant noted that parents are not blamed for giving their children other conditions:

There might be a stigma attached to something you’re carrying around...I don’t know. I just think people are more tolerant of that kind of thing now. We have a neighbor whose son has diabetes and has had since he was a little kid. Nobody thinks, “Well, they gave that to him” (Interview 40).

Indeed, decision making about reproduction and child bearing was among the most frequent concerns raised by participants in this research.

### **Genetics and Reproductive Decision Making**

Although we did not ask participants directly about their beliefs about the appropriate uses of genetic information in decision-making about reproduction and child bearing, this emerged as a strong theme in the interviews. While respondents expressed concerns about eugenics, as described above, they also framed genetic information as something that could help people with epilepsy and their family members make “informed” and “responsible”

decisions about reproduction. However, the meaning of “responsible action” varied among study participants.

Some respondents suggested that if they or their family members knew that they carried a gene associated with epilepsy, they might choose not to have biological children. Speaking for herself, one woman with epilepsy commented that being told she carries a gene for epilepsy “would really strongly influence me towards adopting” (Interview 23). Adoption was also raised by a family member, who commented that genetic information could help people with epilepsy decide

whether they really want to have children or whether they want to go ahead and adopt children so that you know, if it does go from generation to generation you know we’ll just stop it here. And that way any future generations won’t have to worry about it (Interview 2).

Approximately 50% of the participants in this study are 50 or older (with an average age of 48), and more than 80% already have had at least one child. As such, many of their comments about the implications of genetic information for their own reproductive decision making were retrospective. Looking back, respondents speculated that if they had access to genetic information when they were of childbearing age, and were told that they carried a gene for epilepsy, they may have chosen not to have children.

I would have taken it, if I would have heard about it in my 20’s, I would have wanted to take it. And if it did come back positive it would have influenced me whether or not I would have kids, probably to not have children at that time. I remember how I thought about it then. I would have wanted to take the test then. And if it came back positive, I wouldn’t have wanted to have children (Interview 28).

These speculations are clearly influenced by respondents’ experiences of having children with epilepsy, as evident in the comments of this mother:

Epilepsy...sure can make your life hell. In retrospect, if I were going to have children now and if I would know that there’s a way to tell whether or not you’re going to have children that have epilepsy by some kind of study, I’d want to know. If I could do some gene selection *because knowing what I know now...* I mean, we had a pretty rough go of it (Interview 31).

Because women’s reflections on choices made many years ago have been shaped by events following their decisions – which can include the complexities of managing a child’s medical treatment, witnessing a child’s experiences of stigma, and the effects of epilepsy on family relationships – these retrospective comments are not likely to be accurate predictors of the reproductive decision-making processes of women with epilepsy. However, they do highlight an area of concern.

Additionally, many respondents stated clearly that they feared that genetic information would be used to dissuade women with epilepsy from having children or to terminate a pregnancy if they found out that their child would have epilepsy. In most cases, respondents identified these possibilities in response to questions about the “potential harm” of genetic testing:

I think that if people knew that they could pass it on, they may be less willing to have [children], they may be more scared to have children. I think that that can be harmful (Interview 16).

Well, it might discourage them from child-bearing, or you know, having children and it might not even be a problem.... It could maybe alarm you when you didn't need to be alarmed. (Interview 39)

Respondents were particularly uncomfortable with the possibility of prenatal genetic testing for epilepsy, often stating directly that they do not see epilepsy as a “reason to terminate a pregnancy” (Interview 6) and “if they're going to actually abort kids because they have that gene, that's wrong” (Interview 22).

At the same time, there were women in this study who had chosen not to have biological offspring based on their family's history of epilepsy. This respondent recounted telling her husband that given the prevalence of epilepsy in her family

'I think we should not procreate.' I do. I mean I think that-I think that there's so many kids out there that need to be adopted and need good families...and it [epilepsy] is a lot of work and [there's] ...a lot of stigma to work against... (Interview 32)

Another respondent said “If I knew there wasn't a gene I could pass on, I'd have more [children] of my own” (Interview 36). For this subset of women (some of whom did not have epilepsy themselves), being able to obtain accurate information about epilepsy genetics and risk to offspring – whether through genetic testing or through genetic counseling (Helbig et al., 2010) - might increase their sense of their reproductive options.

A big part of what is at stake in reproductive decision making is our understanding of epilepsy itself, as this respondent stated clearly:

I think where I have mixed emotions is if some mom wants to go in and they do an amniocentesis and they check it for that gene... And you know, I understand how parents would want to know in advance whether or not their child would be born with defects and so forth. *I just don't think epilepsy is a defect* (Interview 13, emphasis added).

Similarly, a participant asked rhetorically, “I mean is that the worse thing that can happen to your kid, being an epileptic?” (Interview 24). Indeed, respondents comments highlight concerns about how genetic information will shape understandings of what epilepsy is and what it means to live with this complex condition.

### What Epilepsy Is

Respondents told us directly that they were concerned about how genetic information would interact with “the perception of what epilepsy is” (Interview 13). Their comments highlight how the biological and social specificities of epilepsy may shape what is at stake in the advent of genetic testing for a complex, clinically heterogeneous, and stigmatized condition.

Respondents position the clinical heterogeneity and genetic complexity of epilepsy as reasons for caution in interpreting and making decisions based on genetic information. Especially in their comments about reproductive decision-making, respondents noted the importance of understanding that there are “different degrees or different forms” of epilepsy (Interview 3) and significant variability in epilepsy symptoms, including responses to medication; for example, approximately 60% percent of people with epilepsy can be expected to enter long-term remission upon initiation of treatment, and of those, about 50% will be able to withdraw from medications and remain seizure-free (Kwan & Sander, 2004). As this respondent emphasized:

I would counsel them to not avoid having children. ...This is a very controllable epilepsy. It's not a handicapping type disease process...there is a risk, but I

wouldn't change any life decision based on what we know of this form of epilepsy. There are other genetic diseases where the answer would be different. *But just because it's a genetic disease I wouldn't...suggest that it was an alarming problem.* [It's] a manageable one... (Interview 29, emphasis added)

Related to this, respondents argued that "something in your blood" is not a good measure of what will happen since "there are so many degrees of the illness... [and] there are ways of controlling epilepsy..." (Interview 18). Another respondent stated repeatedly "you can live with epilepsy" (Interview 37). Respondents asserted also that genetic testing is an unreliable basis for reproductive decision making for conditions like epilepsy since "there is only the chance, just because you have the gene, doesn't mean you're going to have it" (Interview 6). For example, a respondent noted that because of the complexity of epilepsy genetics, interpreting prenatal genetic testing is especially problematic:

I mean I'm not anti-abortion in the least, but I think it's a slippery slope of okay, you know, this child-we can tell this fetus has extreme epilepsy...it's got all of these gene(s). But...this fetus may have slight epilepsy. And where's the decision where you abort and whether you'll make those decisions. (Interview 23).

These comments point to concern that genetic testing information might make epilepsy seem more dreadful, less varied, and more genetically determined than it is, thereby constraining appropriately nuanced understandings of the condition among those affected by it.

Conversely, there was a strong sense among respondents that research on epilepsy genetics might improve the lives of people with epilepsy by making it a more well understood and less "scary" condition. Respondents suggested that genetics research might offer a particularly powerful means of increasing public understanding of epilepsy (Interview 32), dispelling myths about seizures (Interview 11), and making it more comfortable for people with epilepsy to disclose their condition to others:

Because then... you take away an element of fear from it. It's not such an unknown. It's not such a scary thing. It's genetic. There's an answer to it...and so if you have this huge press release of genetic testing in relation to epilepsy all of this and people are talking about epilepsy, and just that conversation can just having in the mainstream media is going to be so helpful alleviating people's fear and unknown...I would probably feel much more comfortable telling people, because it wouldn't be an unknown (Interview 16).

In these remarks, people with epilepsy suggest that because the public understands so little about epilepsy, genetic information provides an opportunity for positively transforming public understandings of what epilepsy is.

Despite the difference in emphasis across these two sets of comments, they express a shared belief that part of what is at stake in epilepsy genetics is *how the condition itself is understood*. Taken together, they suggest that understanding the implications of genetic information requires close attention to the biological and social particularities of a condition. Moreover, they highlight the importance of considering how genetic information can be used to either reify or dismantle the labels and stereotypes (Link & Phelan, 2001) associated with stigmatized illnesses.

## Conclusions: What's at Stake?

The goal of this paper was to explore how people with epilepsy and their family members understand epilepsy genetics and perceive the benefits and risks of genetic testing for epilepsy. Because epilepsy historically has been a highly stigmatized condition, we began this project with a particular concern about the effects of genetic testing on stigma.



However, in their responses to open-ended interview questions, study participants also identified a number of novel concerns.

Similar to research on beliefs about genetics among people in families at risk for Alzheimer's disease (Lock et al., 2006), hereditary non-polyposis colon cancer (McAllister, 2003), as well as families who utilize genetic counseling for a wide range of concerns (Chapple et al., 1995), we found that participants in this study hold "personal theories of inheritance" (McAllister, 2003) that deviate from standard scientific models. In particular, respondents expressed belief that shared physical characteristics – such as eye or hair color – and/or personality traits, can predict patterns of epilepsy inheritance in families. This finding is concordant with previous research that suggests that people commonly assume that phenotypic resemblances shared among certain family members predict shared disease risk among those family members (Lock et al., 2006; Richards, 1996).

Beliefs about the heritability of epilepsy have served as a rationale for some of the worst forms of stigma and discrimination possible, including institutionalization, prohibitions on marriage and immigration, and forced sterilization (Schneider & Conrad, 1983; Temkin, 1971). Despite this troubled history, people with epilepsy and their family members believe that genetic information offers myriad potential benefits. The majority of participants in this study say they would undergo genetic testing, provided that adequate protections were in place to address the risks of stigma and discrimination against themselves and their family members.

The possibility that genetic information might serve as a basis for discrimination in employment, health and life insurance is a prominent concern for the people who we interviewed. Fear of stigma and discrimination are not unique to people with epilepsy and their family members. Previous studies have demonstrated that the potential loss of employment, health insurance, and life insurance are major concerns among people considering genetic testing for a variety of conditions (Bombard et al., 2008; Catz et al., 2005; Lynch et al., 1997; Taylor et al., 2008). Concern about the implications of genetic information for family members' has been identified as a reason for declining genetic testing (Phillips et al., 2000). Because this study was conducted before the passage of the Genetic Information Nondiscrimination Act (Asmonga, 2008; Korobkin & Rajkumar, 2008), we could not assess whether and to what extent this policy has alleviated such concerns. This is clearly an important topic for future research.

Our interviews with people with epilepsy and their family members point to a host of concerns that have yet to be addressed either in public policy or guidelines for genetic testing in the epilepsies. First, many respondents expressed concern about the possibility that people who learn that they carry a gene associated with epilepsy might "limit" themselves or feel constrained in their life goals. Second, while we did not ask directly about the use of genetic information in reproductive decision-making, respondents repeatedly raised this as a key issue in their lives and the lives of their family members. Some respondents expressed fear that genetic information might be used to pressure people with epilepsy not to have children; they likewise tended to be critical of the possibility of prenatal genetic testing for epilepsy, which would raise the possibility of selective abortion. Taken together, these comments suggest that questions regarding what epilepsy *is* and how it shapes one's life are part of what is at stake for people with epilepsy and their family members, as they seek to make sense of genetic information about this condition.

On the whole, participants in this study suggested that because of the clinical heterogeneity and genetic complexity of epilepsy, genetic information often will not be a suitable basis for significant life decisions, including whether to have (biological) children. However, since

individuals were included in this study only if they had participated in EFSCU, they are likely to have an unusually nuanced understanding of epilepsy genetics and an exceptionally favorable disposition towards genetic research. These issues will require ongoing attention – in clinics, in research, and in advocacy – as genetic information becomes increasingly available to people with epilepsy and their family members.

Our research demonstrates that engaging with the perspectives of people with epilepsy and their family members is critical to understanding the implications of contemporary epilepsy genetic research and testing. This analysis should inform efforts to develop guidelines to determine the conditions under which testing is conducted, to help people with epilepsy and/or their family members consider the risks and benefits of genetic information, and to assist voluntary organizations in their advocacy and educational campaigns on behalf of people affected by epilepsy.

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### Research Highlights

- Genetics is increasingly a part of the experience and self-understanding of many people with epilepsy and their family members.
- People in families affected by epilepsy have “personal theories of inheritance” regarding what “heredity” and “genetics” mean.
- Many people with epilepsy and their family members assert that they would utilize genetic testing if it were offered to them.
- Genetic information raises concerns about stigma, discrimination, reproductive decision making, and the life chances of people with epilepsy.

**TABLE 1**

Sociodemographic characteristics of study participants

	People with Epilepsy	Family Members	Total
<b>Total participants</b>	22	18	40
<b>Sex</b>			
Female	16	12	28
Male	6	6	12
<b>Marital status</b>			
Married	14	14	28
Single	2	2	4
Living with a partner	2	1	3
Divorced or separated	4	1	5
<b>Age Range</b>			
<18	0	0	0
18–22	1	0	1
23–30	3	0	3
31–40	5	2	7
41–50	3	6	9
>50	10	10	20
average (years)	45.9	50.1	47.8
<b>Education</b>			
High school graduate	3	0	3
Some college	6	4	10
College graduate	8	6	14
Graduate school	4	7	11
Missing Data	1	1	2
<b>Children</b>			
Yes	16	17	33
No	6	1	7
<b>Occupation</b>			
Unemployed	2	0	2
Employed	16	17	33
Retired	3	0	3
Missing data	1	1	2
<b>Household Income (U.S. dollars)</b>			
Under \$20,000	2	0	2
\$20,000 to \$40,000	4	3	7
\$40,000 to \$60,000	2	3	5
\$60,000 to \$80,000	1	4	5
\$80,000 or more	12	5	17

	<b>People with Epilepsy</b>	<b>Family Members</b>	<b>Total</b>
refused	1	1	2
Missing Data	0	2	2