

Science and Suffering: Genetics and the Lived Experience of Illness

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ABSTRACT

With more than 10,000 conditions connected to pathological genes, genetic science has the potential to impact illness experience substantively. Building on medical sociological and science studies literatures, my study analyzes how genetic discourses and technologies shape the lived-experience of Huntington Disease (HD). Analysis draws on in-depth interviews conducted in Canada with 24 individuals with the HD mutation and 14 caregivers (e.g., spouses). Study findings detail how genetic discourses and illness experiences intersect to produce “genetic suffering,” a participant-derived concept describing a novel modality of suffering. Genetic suffering is detailed in relation to four themes: 1) Guilt, responsibility, and genetic inheritance, 2) Chance, uncertainty and genetic testing, 3) Ambiguity and genetic onset, and 4) Fatalism and genetic prognosis. After describing the intersections between the science of genetics and suffering in HD families, I discuss the implications of study findings for debates on genetic responsibility and consider the unintended consequences of genetic technologies.

KEYWORDS: genetics; genetic testing; genetic responsibility; illness experience; Huntington Disease.

The science of genetics¹ has the potential to impact society substantively. Genes have been associated with more than 10,000 conditions, suggesting the possibility of genetic technologies to reshape understandings of health and illness (WHO 2015). Although genetic research currently garners considerable public support, annually receiving over 20% of National Institutes of Health’s (NIH) total research budget (NIH, 2015), the ramifications of genetic technologies for patient populations and society have been hotly debated. Advocates of genetic technologies defend profiling patients via race and ancestry to provide tailored interventions and diagnoses (Hoffman et al. 2015; Staiano,

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1 From here on, I will shorten “science of genetics” to “genetics.”

Bouchard and Katzmarzyk 2013). Advocates also argue that genetics enable individuals to increase their medical knowledge, ameliorating lay-expert divides through the development of the lay expertise of citizens (Novas and Rose 2000; Novas 2007; Rose 2007). In contrast, other scholars have critiqued applications of genetic science (Conrad 1997; Duster 2003; Fujimura et al. 2014; Rafter 1992; Rothman 1995), linking it to increased medicalization (Conrad 2005), increased fatalism (Hallowell 1999) and selective reproduction (Rapp 1999; Timmermans and Buchbinder 2013).

My analysis engages with the medical genetics component of this debate, investigating how genetic discourses and technologies impact the lived-experience of illness in Huntington Disease (HD). Given the breadth of genetics, I focus on four aspects of genetic discourse and technology in the context of the study of disease. Specifically, I analyze participants' conceptualizations of heredity (*Guilt, Responsibility, and Genetic Inheritance*), pre-symptomatic testing (*Chance, Uncertainty, and Genetic Testing*), disease trajectory (*Ambiguity and Genetic Onset*) and prognosis (*Fatalism and Genetic Prognosis*). These four themes constitute what I term "genetic suffering," a participant derived concept describing an intersection between science and suffering, or how conceptualizations of genetic science and technology are incorporated into the lived-experience of illness, facilitating a unique modality of suffering distinct from HD symptoms. Contrary to advocates (Rose 2007) and aligned with critical perspectives of genetics (Hallowell 1999), I argue that a novel vector of suffering is an unintended consequence of participants' encounters with genetic discourses and technologies. Simply stated, as I show in the data below, participants suffered *with* science, with their perceptions of genetic discourses and technologies woven into distressing encounters with HD. While I explicitly focus on heredity, pre-symptomatic testing, disease trajectory and prognosis in the context of HD, I also discuss findings in relation to other genetic conditions.

Huntington Disease provides a strong case to examine intersections between genetics and illness experience. HD comprehensively impacts an individual, causing dance-like motor movements, impairing cognitive functioning, altering mood, and creating personality changes. HD has also featured extensively in the history of genetics, including the identification of the hereditary inheritance of HD, its role in eugenics campaigns, and the development of one of the first genetic screens for a disease (Davenport and Muncy 1916; Huntington 1967/1872; Wexler 1995). Furthermore, the number of studies examining the impact of genetic testing for HD is "unrivaled" compared to other genetic conditions, with the majority of studies concluding the impacts are positive (Paulsen et al. 2013, p.7). Considering the impact of HD on health and well-being, its complex history, and the predominately positive assessments of the impact of genetic technologies, this paper investigates the relationship between lived experience of illness and the ramifications of genetic technologies in the context of HD.

This paper has three aims. First, I provide an analysis of the lived-experience of illness of HD, an illness that receives scant sociological attention despite being one of the first conditions with pre-symptomatic genetic testing, a target of early eugenics campaigns, a source of severe stigma, and a condition that causes comprehensive suffering. Second, I respond to Casper and Morrison's (2010) call for increased dialogue between medical sociology and science studies by drawing on medical sociological research on the lived-experience of illness (Bury 1982; Charmaz 1991) and science studies' analyses of the interplay between science, categories and subjectivities (Hacking 1995; Mol 2002) to analyze an encounter between genetics and the lived-experience of HD. Here, as discussed above, I contribute the concept of "genetic suffering." Third, I consider debates on genetics in relation to HD families' experiences. Specifically, in the context of genetic knowledge of disease, I argue that genetic discourses and genetic technologies (e.g., heredity, pre-symptomatic testing) have unintended consequences for patient populations, which I connect to a critical commentary on genetic responsibility.

HUNTINGTON DISEASE: HISTORY AND EXPERIENCES

Huntington Disease was initially associated with demonic possession, mental illness and various types of chorea (Hayden 1981) before George Huntington's (1967/1872) description of the disease, it's

symptoms and autosomal dominant inheritance pattern. Davenport and Muncey (1916) subsequently cited this inheritance pattern to justify forced sterilization and surveillance. Wexler (2010) argued that such eugenics campaigns exacerbated the stigmatization of HD, which persists to the present day (Bombard et al. 2012). HD is currently estimated to affect 12-14 people per 100,000 (Fisher 2012), with an additional 20 people, such as spouses and friends, impacted by each diagnosis (Aubeeluck and Moskowitz 2008).

In the 1980s, genetic markers for HD were discovered that permitted an early version of pre-symptomatic genetic testing, with definitive genetic testing becoming available in 1993 (Adam et al. 1995; Wexler 1995). This genetic test was one of the first for any condition and can determine whether an individual has the trinucleotide repeat expansion (cytosine-adenine-guanine [CAG]) that causes HD. Those with one affected parent are considered at-risk and are eligible for genetic testing once they reach the age of majority, although they typically must undergo genetic counseling beforehand (Nance et al. 2011). On occasion, minors can receive testing with a medically compelling reason (HDNY.org 2014).

Aside from a gene-negative result, testing has several potential outcomes. Individuals with a repeat of 27 to 35 will not develop HD, although their children might develop HD through meiotic instability (Nance et al. 2011). Individuals with repeats between 36 and 39 are intermediate cases, and might or might not develop HD (Semaka, Balneaves and Hayden 2013). Individuals with repeats greater than 40 will develop HD symptoms during a typical lifespan (Nance et al. 2011; Semaka et al. 2013). Juvenile HD, where symptoms manifest in one's early twenties, occurs with repeats of 60 or higher, although it has been observed in repeats of 45-58 (Biglan and Shoulson 2007). At the population-level, there is an inverse relationship between CAG-length and age of onset; however, this relationship is not consistent enough to predict onset age or symptom severity for an individual (Nance et al. 2011).

For the majority of people, symptom onset begins in mid-life. Initial symptoms are often cognitive or psychiatric, such as disruptions to memory, inability to multi-task, mood changes and hallucinations (Nance et al. 2011). However, HD is perhaps most associated with its chorea or involuntary dance-like movements throughout the body. The disease is degenerative, and symptom severity escalates until individuals require managed care. The disease is fatal and individuals typically succumb 15 to 20 years after symptom onset (Walker 2007).

The development of genetic testing has profoundly impacted HD families. Given the repercussions of a positive test, people with HD have reported difficulty communicating with family members and medical professionals about genetic risk and status, while some have also reported feeling pressured by family members to undergo testing (Etchegary 2009; Forrest et al. 2003; Taylor 2005). Studies have also identified the ramifications of a negative test (Huggins et al. 1992), with 10% of individuals testing negative experiencing "severe difficulties with coping to their new [genetic] status" (Wiggins et al. 1992, p. 1405). Similarly, recent research suggests that individuals with intermediate repeats (36-39) might struggle to understand HD's impact on their lives and reproductive decisions (Semaka et al. 2013). Members of HD families, regardless of whether they are gene-positive, symptomatic or at-risk, have also reported experiences of genetic discrimination from insurance companies and employers (Bombard et al. 2012). However, a recent review of HD pre-symptomatic testing concluded that testing predominately improved quality-of-life (Paulsen et al. 2013), with only four of 28 studies reporting negative repercussions for mutation carriers and most differences between those testing positive and negative "dissipat[ing] with time" (p. 7).

Social scientists have also examined HD. In relation to lay knowledge and genetic discourses, Richards (1993) argued benefits of genetic testing might not be salient to HD families, as individual identity could be difficult to reconcile with test results. Similarly, drawing on interviews with 41 HD family members, Cox and McKellin (1999) reported that lay constructions of genetic risk are social, fluid and can contradict scientific perspectives. For example, their participants equated geographic and social proximity to gene-positive family members to genetic proximity in estimating genetic risk.

Cox (2003) also detailed lay narratives on the decision to undergo testing, arguing that family experience with HD can influence how individuals arrive at a testing decision.

Other consequences of genetic testing have also been explored. In a study of 26 HD families, Konrad (2005) described how receiving a positive test might create “pre-symptomatic” persons and initiate ethical dilemmas around disclosing genetic status and reproductive choices. Huniche (2011), drawing from interviews with 21 members of HD families and observations of 50 genetic counseling sessions, argued that social scientists need to emphasize the moral legitimacy of declining HD testing. Leontini (2010), analyzing 11 narratives of individuals from HD families, documented ambivalence and resistance in relation to a meta-narrative that individuals have a right to healthy children and that health is about being risk-free. Novas and Rose (2000), who analyzed HD web forums, have argued that, rather than generating experiences of fatalism or narratives of biological reductionism, genetics facilitates the construction of an autonomous and responsible new subject and provides opportunities for building communities that share common experiences.

ILLNESS EXPERIENCE AND SCIENCE

Illness experience research articulates how individuals live with a condition. For example, Frank's (1995) work demonstrated how ill individuals' narratives contrast and contradict with health professionals' accounts, while Williams (1984) detailed how individuals reconstruct narratives to regain stability post-illness. Likewise, Charmaz (1991) documented how chronically ill individuals adjust to loss, deal with isolation, and manage their everyday lives, while Bury (1982) illustrated how illness creates biographical disruptions that alter social relations and perceptions of meaning. Taken together, this research directs analyses to the complexity of the lived-experience of illness and contrasting experiential and allopathic accounts.

Illness experience intersects with multiple social phenomena (Kleinman 1988; Williams 1984). One such intersection, which tends to be treated provisionally in analyses of illness experience, is that with science. Scientific developments have numerous potential ramifications for illness experience. For instance, Mol (2002) described how atherosclerosis is “done” differently by various medical professions, such that different professional groups produce distinctly different and mutually exclusive versions of disease. Similarly, Hacking (1995) demonstrated how scientific conceptualizations impact the experience of psychiatric disorder. Specifically, Hacking detailed how the actions of patients diagnosed with multiple personality disorder led to changes in the definition of the disorder, and, in turn, how mental health professionals' elicited “alter” personalities. This work emphasizes potential analytic connections between scientific conceptualizations and illness experiences.

Existing research on how genetic conceptualizations and practices might impact illness reveals contradictory findings (Phelan 2005; Easter 2012). Central to this issue are debates over genetic risk and responsibility. For instance, Novas and Rose (2000) argued that “far from generating fatalism,” (p.485) genetic risk facilitates a new form of genetic responsibility, which facilitates autonomy, self-actualization, and levels doctor-patient interactions. Other scholars have countered that genetic responsibility can precipitate blame within families and communities (Arribas-Ayllon, Sarangi and Clarke 2008; Mozersky 2012), facilitate selective reproduction (Rapp 1999; Timmermans and Buchbinder 2013) and limit patients' medical choices (Hallowell 1999). This debate directs analyses to the ramifications of genetic discourses for illness experience.

The relationship between lay and scientific knowledge is central to the interplay between genetic discourses and lived-experience. Richards and Ponder (1996) documented that individuals consistently underestimate their degree of genetic connection to non-parental family members, while Richards (1993) argued that there is a disjuncture between medical professionals' and lay individuals' perceptions of risk, screening and inheritance, with at-risk individuals over-estimating their likelihood of developing diseases. Additionally, Condit (1999) observed that lay individuals endorse non-deterministic interpretations of genetic science and further (Condit, Parrot and Harris 2006)

suggested that laypersons perceive individual will as a strong causal force independent of genetics. As with the lived-experience literature, this research guides analyses to consider tensions between lay and scientific accounts.

DATA AND METHODS

Thirty-eight people were interviewed for the study. Twenty-four interviewees (13 male; 11 female) had tested positive for the HD gene, with all but four having confirmed symptom onset. The duration between an individual's discovery of HD in their family and interview date ranged from 2 months to 14 years. Ten participants were the first person diagnosed in their family. Symptoms ranged extensively, from four asymptomatic individuals, to two participants requiring managed care. The remaining 14 participants were caregivers (4 male; 10 female). Eleven of the caregivers were spouses, one was a mother and the other a grandmother. All interviews were conducted in Canada.

Data collection spanned seven years (2008-2015). I conducted follow-up interviews with two caregivers and three individuals with HD who were interviewed earlier in the project. The majority of individuals from early data collection had passed away, transitioned to managed care, or were unreachable, leaving me unable to complete additional follow-up interviews. Accordingly, I situate my study design as cross-sectional rather than longitudinal, although I draw on follow-up interviews to provide insights on later HD stages.

I employed maximum variation sampling (Miles and Huberman 1994), a form of purposive sampling that aims to increase variation between interviewees. Specifically, I sampled individuals who were gene-positive and individuals with confirmed HD onset. Furthermore, I sampled individuals with symptoms ranging from minor to pronounced. I also sampled for variation vis-à-vis gender, age of onset, and place of residence (i.e., urban, rural). Recruitment was aided by a social worker who distributed my contact information at HD support groups. I also employed network sampling to recruit individuals who did not attend support meetings. Recruitment was also facilitated by a presentation at an HD society meeting and two volunteer opportunities at an HD retreat.

In-depth, semi-structured interviews were conducted in person (N=36) or over telephone (N=7). All interviews were digitally recorded and ranged from 30 to 150 minutes in duration. Interview questions included: "Recount for me the day you found out you were ill," "What were your thoughts on being diagnosed?," "Are genetic diseases different from other diseases? If so, how?," "What symptoms were you experiencing at this time (if any)?," "What changes have you noticed since your diagnosis?," and "Have you made any preparations for the last stages of your illness?." While study data was partially drawn from these questions, the issue of living with a genetic condition was salient throughout interviews. The study received Institutional Review Board approval and all participants were assigned a pseudonym.

Analysis was guided by grounded theory (Charmaz 2006). The concept of "genetic suffering" first appeared as an *in vivo* code and was developed into the four analytic themes over repeated coding sessions. Thematic analysis was complemented by case-comparison analysis (Miles and Huberman 1994). Specifically, I used a spreadsheet to examine each theme (e.g., *Chance, Uncertainty and Genetic Testing*) in relation to each participant and participant subgroups (e.g., males and females) to ensure findings were not limited to specific interviewees. For instance, as described below (*Fatalism and Genetic Prognosis*), comparing perspectives on healthy behaviors revealed that males tended to maintain unhealthy activities (e.g., smoking).

FINDINGS

Participants described how the "genetic nature" of HD deeply affected their perceptions of the illness. Delores, a 34-year-old healthcare professional, and mother of two, concisely described how genetic discourses shaped the lived-experience of HD. She connected suffering and genetics throughout our interview. When I asked why she used the term "suffering" she stated, "I think it [genetics] is

suffering! I do, I think it's a form of suffering. It's emotional and mental suffering to have that burden. . . I mean, telling my parents (pause) my dad cried because he feels the weight of passing down the gene to me." She continued, arguing that HD genetics was a "terrible weight" that families "had to carry."

Other participants echoed Delores' account. For instance, Gerald stated that he "went into a deep depression" and "began self imploding" after discovering he had a genetic condition, which he likened to receiving a "death sentence." Likewise, Beth, a caregiver, argued that amyotrophic lateral sclerosis, in comparison to HD, "is a much kinder disease" because "it's not genetic." Abigail, a single mother, further emphasized the connection between genetics and suffering:

It's not the dying that bothers me. It's the suffering. It's the living horrifically in my own shit and the quality of life that's not there. It's the being a horrific asshole to my family that upsets me. Being a burden to people, that upsets me. . . . So for me, that's the biggest thing, losing who I know and who identify with as Abigail. My compassion. My love. My fearlessness. My strength. My weaknesses. . . [losing that] is my biggest fear when I read about HD and I was like "how the fuck am I going to swallow that? It takes your perception? Are you fucking kidding me? What fucking disease takes your perception?!" That's just fucking bullshit. And it is what it is. It will manifest how it manifests. You can't change the science. The genetics. It really is in my brain.

Abigail contrasted her ability to handle her death with the distressing certainty of her slow, degenerative decline, derived from her conceptualization of genetics.

Building on participants' accounts, I describe how encounters with genetic discourses and technologies shape illness experience and facilitate the experience of "genetic suffering," which compounded the emotional, physical and cognitive symptoms of HD. I present four themes of "genetic suffering": 1) Guilt, responsibility, and genetic inheritance, 2) Chance, uncertainty, and genetic testing, 3) Ambiguity and genetic onset, and 4) Fatalism and genetic prognosis.

Guilt, Responsibility, and Genetic Inheritance

All participants became aware of their genetic risk or status after they or a family member underwent genetic testing. For instance, Abigail suspected HD after noticing her hospitalized father's symptoms resembled those of her grandfather's. After months of persuasion, her father's physicians completed a genetic test that confirmed her suspicions. Several participants and their family members were also initially misdiagnosed with other, typically psychiatric, disorders (Halpin 2011). Accordingly, participants were often unaware that an autosomal dominant and fatal illness had passed through their family for generations.

Participants described the distress and devastation that occurred when HD was first discovered in their family. Regardless of age, health or any other attribute, the appearance of HD in one family member implicated the others:

It started with my father. We started noticing a change in his behaviors several years ago and it took about two years to get a final diagnosis that it was HD. . . . The part that really gets me and gets people about HD is that it's 50/50 and how it can run through an entire family: aunts, uncles, sisters, brothers. That was one of the biggest things that I had to come to grips with when my father was initially diagnosed. We weren't just talking about my father, now we're talking about my sisters. Now we're talking about my children, my sister's children. And what about all my aunts and uncles, and what about all of my cousins? Oh my goodness! . . . And that was the hardest thing and that was what I think people don't understand is that is the whole ramification of this disease. That it can potentially can be so widespread within the family (Cecilia).

Cecilia, a 42-year-old human resources manager, described an experience common amongst participants. One family member received a genetic test for HD, typically in an attempt to diagnose unexplained symptoms. The test subsequently explained generations of peculiar symptoms and deaths, while questioning the genetic status of siblings, children and grandchildren. While Cecilia described HD “run[ning] through families,” other participants referred to the initial diagnosis as a “flood” or a “bomb” that eliminated huge swaths of their family tree.

Passing the HD gene to offspring, or placing them at genetic risk, was a difficult experience for participants. Kenneth, a 76-year-old computer technician, described calling each of his four children after learning of his diagnosis and apologizing profusely, feeling guilty that his children “now [had] this burden to carry.” Another participant described the difficulty in informing her daughter that she was at-risk for HD, a process that became increasingly stressful when her daughter decided to not disclose her HD risk to her husband, relocated to another country and broke off contact with her family. Feelings of guilt and responsibility impacted family members regardless of genetic risk. In a genetic version of survivor’s guilt, unaffected partners and parents suffered from being free of the genetic risk and physiological effects of HD, while simultaneously witnessing its devastating effects on spouses and children. Sabrina, a 53-year-old mother of two, stated, “I have concern for everyone and a fair amount of guilt. I can honestly say because, in the whole family situation, I know that I am the only one that absolutely will never have it.” The suffering and guilt from genetic diseases was not localized within an individual but, like genetic conditions themselves, was distributed across bodies, families and generations.

In lieu of treatment or a cure for HD, feelings of guilt and responsibility translated into advocacy for self-regulated reproduction. That is, given that participants felt accountable for their DNA and believed the suffering their genes caused could only be addressed via genetic means, they argued that HD should be eradicated by selecting HD-free offspring. Although participants were not explicitly asked about reproductive technologies, they often discussed the genetic risk encountered by their children. For instance, Albert, a 57-year-old former business owner, described the guilt over his son inheriting “his” HD gene. While genetic testing was unavailable when his son was born, Albert discussed how such knowledge would have informed his decision-making:

I’ll be blunt about it, you know. I can tell my son is not going to have a very nice life. He can’t get a date. Girls are not really romantically interested in him and so I feel bad for him about that. And he desperately wants that. And that makes me sad. And had I known this, that I would be causing this, I would have wanted not to have him. But I love him.

Another participant, Samuel, who planned on using reproductive technologies, stated, “I don’t want to bring any more children with [HD]. I want healthy ones, I don’t want to be facing them, ten or fifteen years down the road and see them going through the same thing.” Likewise, Beth, a mother of two at-risk children, argued that selective reproduction was the only way to deal with HD and its terrible consequences:

The only way you’re going to stop this is to not create these children. Who would ever wish that on your child? I mean, a 50/50 chance? That’s a pretty high chance. And if you see what happens to people who have Huntington’s, you would never want anyone to have that. I can’t imagine anything worse.

Beth’s husband was severely incapacitated by HD and was unable to communicate beyond one or two-word sentences. Watching him suffer, she became deeply concerned about her children’s genetic status, informing her perspective on reproduction.

Although participants felt responsible for their children’s genetic status, their ability to make reproductive decisions was significantly constrained, as all the participants with children ($N=33$) had

reproduced before they became aware of HD in their family and only three participants were born after the advent of genetic testing. Furthermore, it is estimated that a quarter to a third of individuals are unaware that they carry the gene (Creighton et al. 2003). While Kenneth, Albert and Samuel might have felt personally responsible for their children's genetic risk, they had little recourse to learn of their own genetic status before reproducing. Therefore, genetic inheritance created a two-fold process of distress, impacting participants both when they were at-risk and again when their own positive test implicated offspring.

Chance, Uncertainty, and Genetic Testing

Participants struggled with the chance they had inherited HD before receiving genetic testing. They described a distressing uncertainty created by the 50/50 odds that they had either inherited a genetic disease or were biomedically unaffected. Participants drew on lay genetic knowledge and information about the test results of other family members to calculate their own genetic risk prior to testing. After testing, participants used their CAG-repeat length to predict how HD might unfold throughout their lives.

Participants often metaphorically expressed the 50/50 odds of inheritance in terms of a card game or spin of a roulette wheel, with equal odds of hitting the jackpot or going bust. For some, the odds were further contextualized by a family members' previous testing results. In a genetic version of the gambler's fallacy, some believed that several prior positive results in the family indicated that their own test would be negative and went into their test hoping to "break the streak." Arthur was optimistic about his chances, "I was playing the odds maker because my sister having had it. I thought the chances of two in one family were not that much." When his test came back positive, he and his wife spent the evening crying in their car before beginning their journey home. Delores also perceived testing as a game of chance:

I really felt I was going to be fine because I'm thinking like "you go in there, and it's 50/50." So you think if you don't find it, it's okay. You can just put it off your plate. You can walk out and just have that freedom of thinking "I don't have to think about this again." And then again if you do have it, then you have this whole thing where you have to deal with it. And it's kind of like a lottery.

Metaphors shape how we interpret and make sense of the world (Lakoff and Johnson, 1980) and have been connected to how individuals locate their hereditary risk for HD (Cox and McKellin 1999). Similarly, participants' gambling metaphor communicates the extreme stakes and uncertainty of testing. For instance, if Delores "won," she could "just have that freedom" of a life without HD; if she lost, she had to deal with a fatal genetic illness, which other participants likened to a "death sentence" or "doomsday." The metaphor also emphasizes participants' feelings of decreased agency, with the test positioned as determining their genetic and medical future. That is, although individuals receiving testing already do or do not have the gene, participants metaphorically positioned the test as an arbiter of future health.

The CAG-repeat length that causes HD, which the majority of participants received after testing, was also used as an indicator of future health. Although repeat length is correlated with symptom severity and onset age at the population level, the correlation is not stable enough for individual-level predictions (Nance et al. 2011). Here, participants' interpretations of repeat length are not reflective of official genetic discourses but indicative of lay interpretation of such discourses (Richards 1993).

Participants used CAG-repeat length to predict disease course and severity by comparing their number to others, or situating themselves as an "intermediate case," meaning they might not ever develop pronounced HD symptoms. For instance, Keith, a 55-year-old trucker, whose CAG-repeat was

at the upper limit of the intermediate range², compared his prognosis to a younger man in his support group, “we had one guy [Jeffrey] at the support group who was like 21 [years-old] or something and he has a caregiver already. My number after I was tested for HD was 41. Well, he’s in the 120 range.” Keith continued that, based on the CAG-repeat discrepancy, he expected his disease trajectory to be much different. The unstable relationship between CAG-repeat, symptom severity and onset was demonstrated by Keith and Jeffrey’s later experiences with HD. When I visited Keith and Jeffrey a few years later, both were living under managed care, largely unable to move or speak. The course of HD could be ambiguous and difficult to predict, varying from individual-to-individual despite observed similarities and differences in CAG-repeat count, with the number providing inadequate predictive certainty.

For some participants, their CAG-repeat was discordant with their HD experience. Kate believed she had a small repeat and was certain she would express only mild HD symptoms. She often socialized with care professionals, participated in advocacy events, and provided guidance to “more symptomatic” individuals. Although she was aware that she had the HD-gene, Kate had not received her CAG count. She eventually became curious and learned her CAG-repeat from her genetic counselor. Kate was dismayed when her results indicated that she was well beyond the intermediate range and would likely develop severe HD symptoms.

Despite the predictive meaning assigned to repeat length, some participants observed that the connection between CAG count and disease severity could be quite arbitrary:

I have four daughters. One of them has been diagnosed at age 45. And she has the same CAG repeat that I have, which is 39. I’m 76 and I’m still functioning quite well, yet here she is, the same CAG and everything, and she just fell apart at 45 (Kenneth).

Kenneth perceived himself as largely unaffected by HD; he did not claim disability benefits and believed that any of his health issues resulted from normal aging. His hope was that his daughter, who had the same CAG-repeat, would experience a similarly mild form of HD. Instead, she became substantially more symptomatic.

Participants used repeat-length to navigate the uncertainty of HD. While the gambling metaphor emphasized participants’ feelings of lost control, CAG-repeat length provided a seemingly objective numeric marker that could be employed to discern the course of HD. The disjuncture between medical genetics (Nance et al. 2011) and participant conceptualizations of the predictive utility of CAG-repeat length are also suggestive of an asymmetry between lay and official genetic discourses (Richards 1993).

Ambiguity and Genetic Onset

Those who inherited the HD-gene confronted a paradox: they would develop a disease during their lifetime, yet they might remain healthy for decades following genetic testing. Most participants spent years in an ambiguous position between health and illness, waiting to transition from “gene-positive” to “symptomatic.” I draw on Kleinman’s (1988) distinction between “illness” (the personal experiences of the sick) and “disease” (the biological features of a condition and its impact on physiology) to analyze this ambiguity.

Gene-positive participants were considered healthy until a neurologist diagnosed HD symptom-onset. Although asymptomatic individuals were considered healthy, or to not yet have a “disease,” many struggled with “illness,” particularly in reference the constant anticipation of symptom onset. Participants vigorously analyzed every forgotten name, dropped glass or facial twitch in an attempt to differentiate mundane accidents from HD onset. Samuel, a dentist, carefully observed his fine-motor skills, continually assessing whether or not he had the ability to maintain his practice. He eventually

2 At the time of the interview, Keith believed his CAG repeat to be in the intermediate range.

lost the necessary deftness and, despite his self-monitoring, co-workers encouraged him into early retirement. Similarly, Albert and Mark, both of whom held corporate positions, scrutinized their business acumen. Albert decided to retire when he believed he was losing his ability to operate spreadsheet software and mentally calculate sales figures. Mark, who was concerned about his ability to remember client details, was “encouraged to retire” just after the 2008 economic collapse. While only Albert voluntarily left employment, all three stated that once routine mistakes became imbued with new significance after learning of their genetic status.

Participants who were asymptomatic, or presymptomatically symptomatic in Konrad’s (2005) terms, echoed the retrospective claims made by Arthur, Albert and Mark:

I took the test and I found out that I was gene-positive. Now I’m always wondering every time I do something “is this the onset?” Like “oh, I forgot somebody’s name” or “oh I dropped something” or “I can’t remember how to do this particular formula.” Just thinking “oh, here we go.” I’ll do something and I’ll think “oh!” Then I sit there and just think “this is silly” and I have to remind myself, “no, you’ve always had a problem remembering this”, or “you are klutzy to begin with, so you can’t say that’s what it is” (Cecelia).

Here, Cecilia described how the ambiguous period before symptom onset was rife with self-doubt, self-policing and anxiety. Cecilia added that pre-symptomatic HD was “very hard for other people to understand. They see me and as we go through life they say, ‘oh, she’s the same as she was two days ago, five years ago, ten years ago’ that’s when they say ‘yeah, it is an illness, but it’s not there.’ And I think they forget.” Participants like Cecilia suffered in anticipation of physiological symptoms, which created genetic “illness” in the absence of genetic “disease.”

The distress caused by the ambiguous disjuncture between genetic “illness” and “disease” was further emphasized by the relief participants felt after symptom-onset was confirmed. Participants stated they felt freed from the “heavy burden” of self-surveillance, even though they would now experience the degenerating effects of HD:

I knew I had the gene, but when it would start and when it wouldn’t start was a lot harder for me to do ‘cause I didn’t know exactly how long and that kind of thing. So once it did officially start then I was like “okay, perfect”, I can be like – “yes, I have this disease”, it’s showing symptoms and that kind of thing and it’s a lot easier, I think, to talk about and tell people (Phyllis).

The distress Phyllis felt while asymptomatic was challenging even to verbalize and, like Cecilia, she found it difficult to explain to friends that she was “sick,” but was “not actually sick right now.” Once symptom-onset aligned “illness” and “disease”, she was able to communicate to those around her that she “actually had something.”

Similarly, Louis, a 60-year-old retired construction worker, stated that he was “relieved” when symptom-onset was confirmed and he “finally knew the unknown” date when HD would begin. In contrast to the ambiguity of the pre-symptomatic stage, he said, “I knew [HD onset] was bad, but at least I could deal with it.” Louis was so relieved that he celebrated symptom-onset by taking his wife on a weekend getaway. His celebratory mood was contrastive to his wife’s perspective, who was “devastated” by his diagnosis and felt like the “whole world [came] crashing down.” The asymmetry between Louis and his wife emphasize the personal and experiential quality of the suffering created by the disjuncture between “illness” and “disease.”

Descriptions’ of symptom-onset as “perfect” do not suggest that participants misunderstood the severity of HD. For instance, Phyllis described how difficult it was to watch her mother succumb from HD, while Louis was deeply concerned about cognitive decline and his ability to converse about politics, one of his favorite pastimes. Instead, participant descriptions emphasize the suffering created

by the ambiguous position between genetic “illness” and “disease,” or how they experientially suffered from HD in the absence of confirmed symptom-onset. Participants preferred to navigate, at least in the short-term, the physiological effects of HD rather than occupy the murky territory between “illness” and “disease.”

FATALISM AND GENETIC PROGNOSIS

Juxtaposed to participants’ feelings of uncertainty and ambiguity were feelings of fatalism. Absent a cure or any means to significantly impede symptom development, participants argued that having a genetic condition decreased agency. Those with non-genetic conditions could improve their health with exercise, eating well, or by avoiding unhealthy practices like smoking and excessive drinking. People with HD, participants argued, would experience the same process of degeneration and palliation regardless of their actions.

Participants emphasized that genetic conditions decreased control, which made them “worse” than their non-genetic counterparts. Peter was concerned that HD made “your mental functions keep decreasing” and “your bodily functions worse;” however, he was particularly distressed by the genetic aspect of HD because “whatever is going to happen *will* happen because it’s a genetic disease” (participant’s emphasis). Here, Peter teased apart the impact of the degenerative course of HD and the genetic etiology of the disease. Specifically, he was not just suffering from the slow escalation of symptoms, which can also be a feature of non-genetic degenerative conditions (e.g. heart disease), but from the genetic basis of the degeneration that, for him, meant that he was completely unable to impact progression. For Peter, the fatalism of having a genetic disease meant that he actively considered taking his own life once symptoms advanced. Keith, a 55-year-old retired truck driver, similarly emphasized the fatalism of HD:

You can prevent a lot. Like diabetes and that kind of thing, like heart disease. You can prevent all those. Watch what you eat and make sure you get a lot of exercise, whereas with something genetic there is not much you can do.

Keith continued by detailing how genetic fatalism affected his health. He was a regular smoker and stated that he had “every intention of quitting” on his 50th birthday but “after the diagnosis I thought ‘why bother?’ and I continued to smoke.” Other participants, mostly men, similarly stated that healthy behaviors felt futile in the context of genetic illness. Like Keith, they continued to smoke and drink excessively, or ceased their attempts to exercise and eat healthy foods. While quality-of-life studies of pre-symptomatic testing suggest few harmful effects (Paulsen et al. 2013), Keith, Peter and other participants’ accounts suggest that some negative ramifications of pre-symptomatic testing remain unaccounted for in current assessments.

In contrast to Keith and Peter, several participants, mostly women, became increasingly engaged in healthy activities after learning they had HD. Grace, a 53-year-old retired nurse, purchased numerous products to improve her health, such as weights and an ergonomic workout machine. She demonstrated the latter to me during our interview, arguing that it helped control her chorea. Similarly, Kate maintained an organic garden inside her apartment and organized vegetarian lunches for care workers and people with HD. However, engagement in healthy activities did not equate to a different perception of fatalism. For instance, after her diagnosis, Delores began to eat better and exercise regularly. She hoped that such actions would have at least a modest impact on her HD but was still distressed by her lack of control:

My friend’s husband is diabetic and he is a raging alcoholic and he smokes like a pack a day. And I think – I could just strangle him ‘cause it’s like – you have diabetes and that sucks. Right? He’s had it from the time he was a kid. But you can do something about it! You can be

healthy! You can control it! You can eat right, you can not drink, you can not smoke, like you can control it to an extent. Like that makes me mad. ‘Cause I look at these people that don’t do anything proactive about their health and it’s – I feel like, here I am doing everything I can, and I’m not in control. That’s hard.

Delores’ frustration about her friend’s inactivity emphasized her perceptions of diminished agency. While she believed her friend could remedy his condition with some hard work and healthy choices, Delores lacked similar opportunities despite her proactive approach to personal health. Like Peter and Keith, she perceived the genetic basis of HD as preventing her from exercising meaningful control.

Contrasting participants’ accounts also suggests a relationship between fatalism and gender, as male and female participants varied vis-à-vis the utility of healthy behaviors. Research suggests that the social construction of masculinity undermine men’s engagement with healthy behaviors (Courtenay 2000). In the context of HD, although the disease is fatal and incurable, research suggests that healthy activities can impact the disease, with diet and exercise prolonging cognitive and motor function, while excessive drinking can exacerbate depression (Nance et al. 2011). Accordingly, gendered health performances might intersect with the experience of genetic suffering, as males and females vary in their response to the perceived fatalism of genetic disease.

DISCUSSION

This analysis demonstrated that encounters with genetic discourses and technologies can shape the lived-experience of illness. My analysis employed the concept “genetic suffering” to describe the impact genetics had on illness experience, in addition to the physical, psychiatric and cognitive symptoms of HD. The four themes that constitute genetic suffering detail the distress associated with inheritance, pre-symptomatic testing, disease trajectory, and prognosis. Although themes overlap and intersect, they were presented in chronological order to loosely correspond with participants’ experiences throughout the life course. For study participants, genetic suffering was a unique modality of illness experience. That is, participants suffered *with* science, with their perceptions of genetic discourses and technologies woven into distressing encounters with HD. For example, descriptions of the inheritance pattern and prognosis of HD (Huntington 1967/1872) were not socially neutral biomedical facts, but were imbued with meaning and intertwined with participants’ experiences of guilt and fatalism. Similarly, pre-symptomatic testing was connected to participants’ distressing experiences with uncertainty vis-à-vis their genetic status and HD symptom onset. Accordingly, following Casper and Morrison (2010), building on science studies (Hacking 1995; Mol 2002) and medical sociological perspectives (Bury 1982; Frank 1995), my analysis suggests that scientific discourses and technologies mediate the lived-experience of illness.

My findings are contrastive with scholarship on genetic responsibility (Novas and Rose 2000; Rose 2007). For instance, Rose and Novas (2003) suggest that genetics “is not taking the form of fatalism” but facilitates a new regime of responsibility and creates a “prudent yet enterprising individual” (p.36), which is not evident from the data presented in this study that instead shows accounts of uncertainty and suffering. Rather than increasing hope and agency, participants’ connected genetics and genetic testing to decreased agency and distressing uncertainty. In particular, there was a substantial degree of anxiety over the “unknown date” of symptom onset and the “lottery” of pre-symptomatic testing. Participants’ accounts also challenge the community-building aspect of genetic responsibility. While an HD diagnosis might unite families, participants argued that it also had implications for health, with HD “run[ing] through entire famil[ies] of aunts, uncles, sisters and brothers,” which, in some instances, contributed to separating families. Furthermore, notions of responsibility were most salient for participants in relation to the extensive guilt they felt for “passing” the HD gene to offspring and statements of moral responsibility connected to their reproductive choices.

Participants' accounts of their reproductive decisions highlight potential disjunctures between the agency and community-building aspects of genetic responsibility, as outlined by [Rose and Novas \(2003\)](#). Specifically, participants' exercise of agency and personal control was connected to individualized burden and personalized guilt. Therefore, my analysis is aligned with previous descriptions of the consequences of genetic responsibility (see [Hallowell 1999](#) for example); while discourses on genetics lead some individuals to embrace notions of "enterprising individuals" and a "moral economy of hope," such narratives cannot be divorced from the intersections between genetics and suffering.

Responding to critics of genetics, [Novas and Rose \(2000\)](#) state they find "little evidence that modern genetic biomedicine dreams of the reduction of the sick person to a passive body-machine that is merely to be the object of a dominating medical expertise" (p.507). While it is difficult to disagree with this claim, it is also an over-simplification of critical studies of genetic science that have instead evaluated the consequences of specific employments of genetics regarding issues such as racial reification ([Fujimura et al. 2014](#)), exploitative data collection ([Fortun 2008](#)), and selective reproduction ([Rapp 1999](#)). Furthermore, while post-eugenic genetic technologies for HD were not created to further "dominating medical expertise" but rather for therapeutic applications, and, ideally, to advance towards a cure, they nonetheless have unintended consequence. Specifically, as I have argued, genetic discourses and technologies facilitated a novel vector of suffering. My analysis, similar to other critiques of genetics, does not equate a dismissal of genetic technologies or invalidation of genetic descriptions of illness, but articulates the pragmatic difficulties that arise for patient populations encountering genetic science. Rather than oversimplifying and dismissing critiques of genetics, it is necessary to seriously evaluate the (un)intended consequences, both positive and negative, of genetic discourses for patient populations.

Study findings are also contrastive with several quality-of-life studies indicative that pre-symptomatic testing for HD does not involve long-term negative consequences and that test-takers generally improve from baseline ([Paulsen et al. 2013](#)). Indeed, while [Dufranse and colleagues \(2011\)](#) suggest that pre-symptomatic testing alleviates uncertainty, a primary component of my analysis was the distressing uncertainty associated with testing. I argue that this discordance in findings is best framed by illness experience literature, which emphasizes the differences between experiential and allopathic accounts of illness (see for example [Frank 1995](#)), with the complexities of the former often omitted from the latter. For instance, while pre-symptomatic testing quality-of-life studies measure psychiatric symptoms and catastrophic events (i.e., suicide), they cannot entirely capture the texture of suffering that is not readily translatable to psychological or psychiatric inventories, such as the communication challenges created by "being sick" but "not actually sick right now." Similarly, measuring how test-takers change from baseline does not capture how test results might inhibit future changes, as detailed in male participants' discussion of fatalism and healthy behaviors. Focusing on pre-symptomatic testing also omits how individuals encounter conceptualizations of heredity, with participants describing a single confirmatory test as a "bomb" or "flood" that impacted the entire family. The discordance between my experiential analysis and quality-of-life assessments indicates the importance of research addressing lived-experience, increasing communication between the social and life sciences, and the need to broaden quality-of-life measures.

While my analysis addressed HD, other studies have documented accounts similar to select themes. In relation to issues of inheritance, [McAllister \(2003\)](#) has described how individuals with colon cancer construct personal theories of inheritance, while [Shostak and colleagues \(2011\)](#) described how people with epilepsy respond to the complex inheritance pattern of the condition. Fatalism related to genetic prognosis has also been documented in other conditions, such as breast cancer ([Hallowell 1999](#)). Distress resulting from the ambiguity of genetic conditions has been analyzed, with researchers detailing feelings of inauthenticity ([Lowton and Gabe 2003](#)), pressure to receive prophylactic surgery ([Press et al. 2005](#)), and challenges associated with being a "patient-in-waiting," ([Timmermans and Buchbinder 2013](#)). This literature and the present study suggest that

genetics might create similar dilemmas for various populations, despite differences in condition. My analysis has contributed to this literature by articulating four dimensions of “genetic suffering,” a potential mid-level concept for future analyses. I have articulated the utility of “genetic suffering” using an autosomal dominant genetic condition; however, much current research aims to identify genomic markers indicative of particular diseases. Although few genomic markers have been identified, future research should consider the intersections between genomics, illness experience and suffering.

Despite the prospective similarities between genetic conditions listed above, my analysis does not propose that the lived-experience of HD generalizes to other conditions or that the four themes presented here exhaust the relevant intersections between genetics and illness experience. Furthermore, ill individuals encounter numerous non-genetic technologies (Joyce 2008; Pickersgill 2011; Shim 2005) that might impact illness experience. There are potentially numerous encounters between scientific discourses, suffering and the lived-experience of illness, suggestive of the need for, and potential of, future research.

While genetic suffering might reflect the lived-experience of individuals with HD and other genetic conditions, my study is limited by its cross-sectional design and use of non-probability sampling. Additional research is necessary, particularly that which applies “genetic suffering” to other conditions, employs representative sampling or uses a longitudinal design. As all participants discovered HD in their family after the advent of genetic testing, findings cannot speak to the experience of individuals who transition into genetic diagnostic regimes. Future comparative research on the intersection of genetics and illness experience should consider the experiences of individuals who transitioned between non-genetic and genetic regimes. Lastly, while my analysis focused on the intersection of genetics and suffering, predominately critiquing arguments on the ability of genetics to facilitate hope, agency and community-building, the differences between these arguments are indicative of the complex relationship between lived-experience and genetic discourses. Future research should remain cognizant of patient populations variegated conceptualizations of genetic discourses and technologies.

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