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Diagnosis, psychiatry and neurology: The case of Huntington Disease

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ABSTRACT

Although Huntington Disease (HD) is recognized as a neurological condition, it has a number of psychiatric effects, with recent studies suggesting that these effects can appear years prior to the telltale neurological symptoms. This trajectory has, in part, led to the misdiagnosis of HD as a psychiatric illness, as explicated in numerous case studies. This paper utilizes HD as a case study to investigate the social consequences of diagnosis by highlighting the tensions and ambiguities between neurology and psychiatry, while also discussing the difficulties that HD creates for psychiatry's diagnostic schema. Findings are based on 30 in-depth interviews conducted with both individuals with HD and informal caregivers (e.g., spouses) in British Columbia, Canada. The findings address numerous instances of misdiagnosis and the resulting negative impacts for individual health and well-being. The findings are further discussed in relation to the work of Bakhtin and Latour, with suggestions presented to ameliorate such misdiagnoses.

Introduction

Huntington Disease (HD) is a hereditary genetic condition that impacts movement, affect and cognition (Paulsen, 2004). The disease is autosomal dominant, meaning that if one parent has the mutation, each child has a 50% chance of inheritance. The illness usually affects adults between the ages of 30 and 45, though symptoms can appear in children and young adults (Paulsen, 2004). Approximately 1 in every 10,000 individuals has HD (Huntington Society of Canada, 2010). A precise genetic test has existed to identify the HD gene mutation since 1993. However, a positive genetic test is not considered a diagnosis of HD, as actual diagnosis can only be made by a neurologist based on motor symptoms and the detection of tangible changes in the individual's brain (Rosenblatt, Ranen, Nance & Paulson, 1999).

Even though HD has a number of symptoms associated with neurological ailments (e.g., chorea), it also has symptoms that may appear indicative of a psychiatric disorder, including mood changes and hallucinations (Paulsen, 2004; Yu, 2004). Recent research also indicates that HD's psychiatric and cognitive changes may appear years before the characteristic neurological changes (e.g., Stout et al., 2007). Although there is genetic testing for HD, the disease's symptomology and progression can make accurate diagnosis a complex process, particularly given the estimated 25% of individuals who are currently unaware that HD is present in their family (Almqvist, Elterman, MacLeod, & Hayden, 2001)

The following manuscript suggests that the interpretation of HD's features within psychiatry's diagnostic schema highlights ambiguities between psychiatry and neurology. While both disciplines study the same organ, they describe it in different terms, with neurologists seeking to uncover structural damage, while psychiatrists focus on mental, emotional and behavioural disorders. However, these ambiguities go beyond simple epistemological differences, as they also have tangible effects, made evident in numerous case studies detailing the psychiatric misdiagnosis of HD (e.g., Yu, 2004). Although these case studies detail the misdiagnosis of one or two individuals from a clinical perspective, no published study has yet engaged with the HD community regarding psychiatric misdiagnosis (i.e., incorrect diagnoses) or the social consequences of diagnosis. This report will address this gap by highlighting the accounts of individuals currently living with HD. Their recollections of both personal and familial misdiagnoses will be used to elucidate the consequences of the disjuncture between neurology and psychiatry, which include grief, confusion, incorrect treatment and the application of misdiagnoses to similarly affected family members.

Findings will be discussed in relation to three theoretical concepts. First, Latour's (2005) concept of "oligopitcons", the numerous finely tuned centres of scientific observation, will be used to discuss the overlapping gazes of neurology and psychiatry. Second, Bakhtin's (1986) notion of speech genres will be used to ground the problem of misdiagnosis within actual, everyday speech, while highlighting misdiagnosis as an issue of

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interdisciplinary ambiguity, rather than a lack of competence amongst specific clinicians. Third, Latour's (1987) notion of metrology, the system of standards by which observations made from different contexts can be translated and interpreted, will be used to discuss psychiatry's diagnostics tools.

Diagnosis, misdiagnosis and psychiatric epistemology

There is a long history of critique of psychiatric diagnoses. Perhaps most famous amongst these is Rosenhan's (1973) article questioning psychiatry's inability to distinguish the sane from the insane through the documented diagnosis and psychiatric hospitalization of several healthy individuals. Other critiques include questions over psychiatry's cross-cultural applicability (Good, 1993), the accuracy of its diagnostic categories (Greenberg, 1977), the repercussions of internal divisions on diagnosis selection (Brown, 1990), the disjuncture between its medical training and in situ experiences (Thomas-MacLean & Stoppard, 2004), and the fabrication and alteration of diagnoses by psychiatrists (Pallone & Hennesy, 1994).

Recent research does, however, indicate that psychiatrists do adapt to external criticisms and are cognizant of the ambiguities of their epistemology. For instance, Messinger (2007) described the negotiated construction of psychiatric diagnoses by an interdisciplinary team, including the solicitation of input from non-psychiatrists in an urban hospital. Additionally, Rafalovich (2005) documented clinicians' reflexivity to external, particularly sociological, critiques of ADHD, as well as clinicians' own concerns about the diagnostic criteria for the condition.

Within the context of this paper, the investigation of psychiatric diagnoses is backgrounded by the numerous difficulties that already occur after a diagnosis of HD. For instance, individuals can face stigma based on their movements (Paulsen, 2004), as well as employment and insurance discrimination over their genetic status (Bombard et al., 2008). The familial and hereditary nature of HD also places the family in a unique situation, as they struggle with communicating genetic risk (e.g., Cox & McKellin, 1999). Thus, an *accurate* diagnosis of HD carries with it major implications for both the individual and their loved ones. Misdiagnoses further delay the onset of encountering these issues, as well as lessening the time individuals can engage with them. Psychiatric misdiagnosis also carries its own consequences, as individuals with psychiatric illness also often face stigmatization (Sayre, 2000) and can develop psychological problems in response to their misdiagnosis (Floyd, 1997).

Neurology, psychiatry, and Huntington Disease

Numerous reports indicate that the psychiatric components of HD can be the first symptoms to emerge (Stout et al., 2007). For instance, Duff, Paulsen, Beglinger, Langbehn, and Stout (2007) Stout's study of 681 individuals with HD suggests that psychiatric symptoms appear well before the cognitive and neurological markers of the illness, with emotional dysfunction often being the first symptom. A report by Stout et al. (2007) also suggests that cognitive decline escalates well before the neurological diagnosis, and, similar to the emotional effects of HD, can be an "advanced warning" of motor symptom onset.

The literature on the early psychiatric effects of HD highlights how the disease can blur the boundaries between "neurological" and "psychiatric" illness. While the disease itself can manifest as a hybrid of neurological and psychiatric symptoms, the disciplines of neurology and psychiatry are not as seamlessly interwoven. Indeed, psychiatrists and neurologists have themselves noted the tension between their two perspectives (e.g., Butler, Corboy, & Filley, 2009; Schon, MacKay, & Fernandez, 2006). A trio of neurologists, Price, Adams, and Coyle (2000), most directly summarized this tension as a "great divide", centering around neurology's tendency to focus on the "brain" compared to psychiatry's tendency to focus on the "mind". Although these disciplines have been similar but separate throughout their histories, it is precisely diseases such as HD that create the need for greater disciplinary interchange, leading to Price et al's (2000) suggestion that these professions require a more harmonious education. Price et al. (2000) and others' focusi on this disjuncture (e.g., Schon et al., 2006) tends to specifically address epistemology and pedagogy. However, these different conceptualizations of the mind/brain have repercussions outside of academia, as made especially evident in the misdiagnosis of HD as a psychiatric illness.

A review of misdiagnosis case studies reveals multiple instances in which psychiatrists have mistaken HD for a mental disorder. For instance, Duesterhus, Schimmelmann, Wittkugel, and Schulte-Markwort (2004) document the case of a severely depressed child, whose diagnosis was informed by a long-standing family history of Major Depressive Disorder. Of particular interest was the child's father's diagnosis with Major Depressive Disorder with Psychotic Features; it was not until his eventual suicide attempt that his HD was detected, subsequently explaining the family's apparent depressive genealogy. This child and his father's condition greatly improved once they and their healthcare providers were made aware of their actual condition. In this case, psychiatrists noted that the hereditary depression was a "red herring", concluding that mental health professionals needed to be more sensitive to HD. Tost, Wendt, Schmitt, Heinz, and Braus (2004) expressed similar sentiments in their report of a man who had been diagnosed six times with four different conditions over the course of seven years before his eventual HD diagnosis. Even though his condition stabilized after the correct diagnosis, he had already taken anti-psychotic medications for multiple years, and had been both homeless and incarcerated numerous times as a result of the misperception of his psychiatric symptoms.

Fitting with these calls for greater HD awareness amongst psychiatrists, Appollonio, Frisoni, Curtò, Trabucchi, and Frattola (1997), in their report of three cases of misdiagnosed HD, suggested that genetic tests and genealogical histories be included in standard operating procedures for individuals with symptoms similar to HD. Yu (2004) echoed these statements, also noting HD's prima face similarity to first break schizophrenia, estimating that psychiatric disturbances account for as much as 25–80% of the first symptoms of HD.

These psychiatric case histories provide ample evidence of the difficulties in diagnosing HD, as well as the tangible consequences of its misdiagnosis. It is also noteworthy that all of these misdiagnoses were made after the development of the genetic test for HD. Although this test is an accurate tool for detecting HD, the clinician first needs to suspect HD as a possible diagnosis. Unfortunately, each of the aforementioned articles echoes statements made in Stewart's (1989) report on the psychiatric misdiagnosis of HD. A decade prior to all of the previously discussed case studies, in a journal focused on both neurology and psychiatry, Stewart outlined the risk of misdiagnosis and forwarded a protocol stating that individuals who meet two of the three HD symptom criteria should have the disease eliminated as a possibility through differential diagnosis. Despite this timely and well placed advice, the lessons from Stewart's report on misdiagnosis apparently did not cross the "great divide", as case studies continue to echo these recommendations with little evidence of change in diagnostic practices.

At this point, two important factors become clear. First, HD has numerous psychiatric symptoms, which might appear prior to the characteristic chorea. Second, given the psychiatric nature of HD, it is quite possible that an affected individual's first point of contact with the healthcare system will be a psychiatrist. As such, there is a risk of an individual being misdiagnosed with a mental disorder, rather than HD. Given the symptom overlap between HD and other

mental illnesses, it is understandable that these errors are made; however, what is surprising is the proliferation of case studies repeating similar accounts of misdiagnosis and similar suggestions to avoid mistakes. Subsequently, it is arguable that the ambiguous nature of HD needs to be communicated to psychiatrists via a better means than journal case studies.

It is here that one must turn to the orientating text of psychiatry and mental health in North America, *The Diagnostic and Statistical Manual of Mental Disorders* (DSM), currently in the revised version of its fourth edition (APA, 2000). The DSM-IV is the text that organizes, describes and standardizes mental illnesses. Given that the DSM-IV is a pivotal text for providing accurate and uniform descriptions of mental pathologies, paired with the literature on HD misdiagnosis, it would seem the logical place for a concise and clear definition of the psychiatric components of HD. The DSM-IV codification of HD (APA, 2000, p.165) provides a number of important and useful criteria; for instance, it highlights all three primary areas of HD (psychiatric, cognitive and motor) while portraying a descriptive account of individuals who might be presenting with HD. Yet the account, titled "Dementia due to Huntington Disease", is primarily included to attune psychiatrists dealing with an individual already diagnosed with HD to the potential for dementia. Though the entry states that emotional and cognitive changes might "herald" the start of HD, it does not indicate that HD can be mistaken for mental disorders with these specific symptoms.

The DSM-IV HD entry does not convey any of the lessons learned by the numerous psychiatrists who have misdiagnosed the disease. Nor is HD listed as a differential diagnosis in any other category, including Major Depressive Disorder or Schizophrenia, despite the numerous case studies (e.g., Yu, 2004) in which HD has been mistaken for one of these pathologies. Perhaps most telling is that HD is described as a "progressive degenerative disease" that affects mood, rather than a psychiatric or neuropsychiatric disease. Given the limited presentation of HD in the DSM-IV, and the omission of any mention of misdiagnosis, it is understandable that psychiatrists have repeated both each other's mistakes and each other's advice.

The limited DSM treatment of HD, the numerous case studies, discussions of the "great divide", and the symptomology of HD itself all illuminate an epistemological gap between neurology and psychiatry. However, omitted from these accounts is a systematic analysis of the consequences that misdiagnoses have for individuals with HD and their families, which will be illustrated in the following analysis of accounts of misdiagnoses.

Method

Participants

After receiving Behavioural Research Ethics Board approval, twenty individuals with the HD gene mutation were recruited for this study, with all but three having previously received the neurological diagnosis confirming symptom onset. The physiological effects of HD ranged extensively, from the three aforementioned asymptomatic participants, to two participants currently residing under managed care. Ages ranged from 23 to 83 ($M = 54$); twelve of the 20 participants were male. Ten caregivers were also recruited for the study. Nine of the caregivers were the partners of individuals with HD (eight wives and one husband), and one was a grandmother. Several caregivers attended to multiple individuals. The ages of the caregivers ranged from 37 to 64 ($M = 54$). Names of persons and locations have been changed to protect anonymity.

Participants were recruited between 2008 and 2009, through both snowball sampling and a Huntington Disease Resource Centre (HRDC) in British Columbia, Canada. The HRDC is affiliated with an HD-specific medical centre, through which individuals receive genetic testing, counselling, treatment, diagnosis and social support. Participants were eligible for the study if they either a) had HD, or had tested positive for the HD-gene or b) were a caregiver to anyone meeting the first criteria.

Data collection

Qualitative, in-depth, semi-structured interviews were the primary source of data (Lofland & Lofland, 1995). The majority of interviews ($N = 26$) occurred in-person and were conducted at a time and place of the participant's choosing. The remaining interviews occurred over the telephone. All interviews were conducted by the author, digitally recorded and transcribed verbatim. Interviews lasted from thirty minutes to two and a half hours. All participants were required to sign a written consent form, explained to them by the author at the time of the interview. Upon completion of the interview, the author took detailed fieldnotes, including descriptions of the interview location, relevant participant behaviour, and researcher–participant interactions (Emerson, Fretz, & Shaw, 1995).

Data analysis

Data analysis took place within the context of a larger study on HD services and issues relating to death and dying. Issues concerning psychiatric misdiagnosis were not an anticipated focus and emerged during the course of multiple interviews. While not a grounded theory project per se, analyses drew on numerous aspects of that methodology. Specifically, data analysis utilized line-by-line analysis, thematic analysis and case comparison. Line-by-line analysis was conducted on each transcript to generate large codes to efficiently organize the data (Strauss & Corbin, 1998). This coding structure was then used to code the data using NVivo8™ qualitative data management software. Coding runs for each major code were generated and were then subsequently read multiple times to develop emergent and inductive themes. Case comparison analysis, utilizing the Microsoft Excel™ spreadsheet program, was also conducted to compare each individual case across the entire participant sample (Miles & Huberman, 1994). This technique allowed the data to speak to the particulars of an individual case, while also representing aggregate trends. The resultant themes form the basis for the results section. Analyses were not guided by any particular theoretical perspective; instead, as suggested by several researchers (e.g., Latour, 2005; Strauss & Corbin, 1998), theories were utilized to advance specific sections of the analysis.

Results: experiences and recollections of misdiagnosis

When neurologists and psychiatrists discuss the overlaps and disjunctures of their respective positions, their focus often remains on the realms of pedagogy and epistemology (e.g., Price et al., 2000). These debates become somewhat more grounded within the psychiatric case history literature, which illuminates the effects of epistemological differences. At the same time, omitted from both perspectives are the consequences that such disciplinary tensions and misdiagnoses have on the individuals to whom they are actually applied. Although no report has thus far highlighted these experiences, the impacts and lingering effects of misdiagnosis becomes evident through the accounts of the HD community.

While relaying stories of their family history of HD, participants in this study frequently recalled cases of familial psychiatric misdiagnosis, as well as highlighting a general culture of HD misdiagnosis in generations past. Participants most commonly expressed both the historical misdiagnosis of HD and the attitude toward the resulting treatment with the phrase "locked up in Thorn Hill". In this statement, Thorn Hill refers to the psychiatric hospital where many individuals with HD were placed when their illness

was incorrectly perceived as being psychiatric. Keith (55, diagnosed in 2003) noted this when he spoke of his father's treatment: "My father died in Thorn Hill in '63, but we thought he had schizophrenia. If you look at the family tree there's a lot of people dying earlier than they should have." Several other participants who had conducted archival family histories recalled similar misdiagnoses. For instance, Kenneth (76, diagnosed in 2003) stated that, "I've done a lot of genealogy and you have to get the records from the psychiatric hospitals because that's where they were all put. Because they were all thought to be crazy."

Often the consequences of previous familial misdiagnoses were most directly felt when individuals learned that generations of mysterious illnesses or bizarre behaviours were, in fact, HD. For instance, one participant compared the discovery of HD in her family to a "flood" that consumed her family tree:

> I had no family history, which as you know is quite odd given it's a genetic thing (laughs). Basically my dad got diagnosed in 2007. He was having some problems with motor skills, but not really chorea, but more so mental issues. He was having a problem keeping a job, etcetera, etcetera, and the last test that they did was for Huntington's. Just to rule it out. And it came back positive. And he's got seven siblings who all have kids and me and my sister have kids, so it was like this flood up through our family tree and now like half of it is gone (Delores, 34, gene positive).

The clearest implication of this account is the shock created by the sudden knowledge of a genetic disease within the family. Prior knowledge of a familial history of HD might have altered reproductive decisions of several of the aforementioned family members. However, as both family members and health professionals believed the family was affected by a psychiatric disorder, this possibility was not considered.

Other families had similar experiences based on their own histories of erroneous diagnosis. Particularly problematic was when a parent's misdiagnosis also formed the basis of a child's subsequent misdiagnosis. Kate (57, diagnosed in 2005) said:

> He [her father] had been under psychiatric care, when my mom and him got separated that's when he had a "nervous breakdown" and that's when he had been going to a psychiatrist. But it was the Huntington's and nobody knew. That's when they put him out to the crazy clinic and he was probably out there for four or five months and they ended up giving him shock treatments to bring him back because he would just sit there. But they had no idea that it was Huntington's. . . and my brother got sick right after my father died and they thought it was depression. Which was what my father was misdiagnosed with as well.

Here, in an unfortunate irony, it is the father's apparent psychiatric condition that is seen as the hereditary predisposition explaining his son's similar behaviours. Following that misdiagnosis, Kate's brother received inaccurate and inadequate treatment, spending several years of his life in extreme poverty. Indeed, it was not until Kate herself was tested for HD that her father and brother's experiences were put into an accurate context, and the latter began to receive proper health coverage and disability benefits.

Two caregivers, Beth (60) and Sabrina (53), recounted similar situations. Beth recalled that clinicians thought her husband's brother "had some kind of mental relapse, some kind of falling apart. And it wasn't diagnosed for some time because there was no family history." Sabrina (53) recounted a similar story about her husband's brother:

> We sort of thought he'd actually experienced a bit of a mental breakdown. And that's what we took it as because he was just a little bit, you know, kind of bizarre in some of his behaviour. You could call it a bit of a breakdown because we didn't think he had ever had those symptoms prior to that time, and we didn't know what they were. So he just kind of became a different person. And then he started to lose interest in looking after himself he ended up becoming homeless and lived in his car and we had to intervene and have him find shelter. But again, we kind of chalked it up to, "wow, I guess he just has really lost it." But there always was this question as to whether their dad had a mental illness. So that's why I guess he [her husband] was diagnosed first.

Sabrina's family and health professionals suspected her brother-in-law was manifesting a hereditary psychiatric disorder, as his father had expressed similar mysterious symptoms. Her husband (Keith) was the first to be diagnosed with HD in 2003, although he was the third person in his immediate family to present symptoms. Keith's brother did not receive appropriate or adequate care, spending most of the last years of his life living on the streets of a metropolitan Canadian city. As Keith succinctly put it, "my brother would probably still be alive if he hadn't gone undiagnosed [with HD]."

Karen (59) recounted similar problems with the lack of recognition of the psychiatric effects of HD. Karen began caring for her granddaughter after her son-in-law developed HD and had a psychotic break. She started noticing her granddaughter's difficulties meeting developmental milestones and began suspecting HD:

> I knew. She had behaviour difficulties, severe learning difficulties, inarticulate speech, awkward gait when she was walking. I knew that's what it was. She wasn't diagnosed until I guess she was 12 or 13. They thought she had ADHD, Tourette's and Oppositional-Defiant Disorder, but now we know it was all HD… If a parent is seeing a problem with a child, and Huntington's is a possibility, then I think it should be the parent's right to get them tested, because then you know what you are dealing with. I mean, the scattered diagnoses that we had for her before that were just that, scattered diagnoses. And the one thing that covered all of this was Huntington's. And if we'd known that in the first place. I mean, we had this poor child in remedial math classes, in Kumon [learning centre], she was going to learning disabilities centres. If we had known it was Huntington's we wouldn't have pushed all that stuff on her and made her unhappy by forcing her to do this ridiculous homework.

Karen struggled to get her granddaughter evaluated for HD, even though the disease seemed an obvious candidate. She was told that, unless her granddaughter had a major medical problem, such as a seizure, she was not eligible for a diagnosis or genetic test due to her age, as individuals must be at least 18 to receive the test in British Columbia. Karen subsequently followed the advice of health professionals, who suggested her granddaughter's condition was actually psychiatric and could be addressed with behavioural modification and learning resources, which Karen later believed to be both unnecessary and ultimately detrimental to her granddaughter's quality of life.

In addition to the consequences of misdiagnosis, participants also described the series of professionals they encountered during the diagnostic process. For instance, Becky (36, diagnosed in 2007) was receiving psychiatric counselling for anxiety and an eating disorder immediately before her diagnosis of HD. At this time, her husband had noticed personality changes, increased introversion and paranoia, as well as gait and speech changes. Although her clinician did not suspect a neurological disease, her family members convinced her to undergo neurological testing, primarily to consider the possibility of early onset Alzheimer's Disease (AD), which her father had been diagnosed with. Upon her visit with the neurologist, HD was almost immediately suggested based on her

gait. This diagnosis turned out to be correct and her father's diagnosis, along with several previous generations of AD diagnoses, were invalidated.

Becky's experiences outline two important points. First, even though her clinician did not misdiagnose her, she experienced several HD-related mental health issues before the onset of the disease was recognized. During this period, her movements and speech were affected significantly enough for her neurologist to suspect HD prior to conducting any formal examinations. However, her psychiatrist neither addressed these symptoms, nor recommended their evaluation by another health professional. Second, Becky's experience also highlights the importance of accurate and timely diagnosis. Soon after her diagnosis, she began to develop hallucinations and delusions, including believing she was being watched by cameras in her ceiling and reacting to invisible intruders. Although these symptoms were fortunately recognized as related to her HD, it is not difficult to imagine what her possible misdiagnoses might have been had she developed these hallucinations pre-diagnosis.

Grace (53, diagnosed in 2005) similarly described the transition from psychiatric to HD-specific care. Grace suspected that she was sick well before her HD diagnosis, and her ex-husband had also long presumed that she had a serious medical condition, as she recalled him stating, "you've got movements and you're always running around. You're hyper and you're going to have a serious disease." Both of their opinions were primarily informed by Grace's history of awkward movements: "I've always dropped stuff. I've always had trouble finding things and the computer part of my brain just wasn't there and that type of thing." In this instance, as with Becky, a neurological symptom actually presented as the first HD feature detected by both the individual and her family. However, it was issues related to her marital difficulties that influenced Grace to seek the help of a psychiatrist, who prescribed her several medications, including a powerful anti-psychotic medication (Haldol) typically used for people with acute psychosis and schizophrenia. Eventually, given her repetitive and awkward movements, both Grace and her psychiatrist surmised that she had Tourette Syndrome, which was partially informed by her son's previous Tourette's diagnosis:

> I don't have any family history of Huntington's so at work [a hospital] they were saying, 'well, you're probably just stressed or depressed'. And there were movements too. So I ended up finding this gal [a psychiatrist] through my family doctor, to talk about my divorce. And she put me down as stressed and depressed and said that I couldn't work. And she thought I had Tourette's. She was trying to find out about movement disorders as well, 'cause my son had Tourette's. And I was having troubles moving so she just thought I should go to a movement disorders assessment. That's when they started suspecting Huntington's Disease.

Although there was no history of HD in Grace's family (her affected father had been misdiagnosed with Obsessive-Compulsive Disorder), during the differential diagnosis process, one of the diseases her psychiatrist eventually thought to rule out was HD, leading to Grace's eventual genetic testing.

These participants underscore the consequences of HD misdiagnosis, such as incorrect treatment and the reapplication of misdiagnoses to similarly affected family members. Additionally, Becky and Grace describe the process of transitioning from misdiagnosis to HD-specific care, and while Becky's psychiatrist did not suspect another condition, Grace's psychiatrist became her primary advocate in receiving a correct diagnosis. Participants' stories also document the numerous pathologies that HD has been mistaken for, such as Tourette's, AD, Depression and Schizophrenia. They additionally describe the transition from misdiagnosis to diagnosis, detailing the numerous professionals involved and how family members, as well as mental health professionals, mistakenly interpreted HD symptoms.

Discussion

Although the problems of misdiagnosis and potential solutions have been forwarded in the numerous psychiatric case studies, these solutions have not led to an amelioration of misdiagnoses. It is at this juncture that social theory, specifically the works of Latour and Bakhtin, can provide helpful insights by elucidating issues regarding the separation, activation and translation of psychiatric epistemology. First, Latour's concept of oligopticons will be used to further trace the disjuncture between neurology and psychiatry. Second, both Bakhtin's notion of speech genres and Latour's concept of metrology will be used to describe problems with psychiatry's operation and to suggest a possible remedy for this epistemological disjuncture.

Neurology and psychiatry study the same object, but in different ways. Here, Latour's concept of oligopticons provides us with an avenue to further explore the connections and separations between the two disciplines. Latour (1987; 2005) describes oligopticons as the numerous centres of observation and calculation operating within society. In contrast to Foucault's (1975) concept of the panopticon, which exerted a complete and unobscured gaze, oligopticons are less comprehensive, but, as Latour (2005) notes, "what they see, they see it well" (p.181). We can understand neurology and psychiatry as constituting two distinct oligopticons, each observing, describing and operating in relation to the brain. Latour's concept would suggest that, even though these disciplines do observe aspects of the brain in great detail, neither of them perceives it in its totality, with neurology focusing on alterations and injury to organic matter, in contrast to psychiatry's observation of changes in mood and personality and mental disorders.

These boundaries can be traced in greater detail by a brief comparison of HD to AD. Both of these diseases are defined as neurological, but in contrast to HD, the DSM-IV entry for AD is a formal diagnostic category (APA, 2000). The entry for AD reviews all the major features of the disease, which are distinctly psychiatric and cognitive in nature. Thus, although the disease is caused by neurological damage (like HD), its effects are entirely psychiatric/cognitive (unlike HD). Accordingly, even though AD, like HD, is a neurological condition with psychiatric/cognitive effects, it does not exhibit the same disjuncture between neurology and psychiatry, as both of these disciplines can account for its features. This disciplinary overlap does not occur with HD, perhaps as a result of its motor symptoms; nevertheless, these motor symptoms do not explain the omission of HD's numerous psychiatric features from the DSM-IV or its sole inclusion as a sub-type of dementia.

While Latour's concept of oligopticons provides us with a better understanding of the related but disconnected relationship between neurology and psychiatry, Bakhtin's (1986) notion of speech genres allows us to better connect psychiatric observations to their effects on patients by focusing on the discursive nature of the misdiagnosis. The act of diagnosis employs specialized language and forms of communication specific to a given medical institution, as demonstrated by psychiatrists' misdiagnosis of HD as a psychiatric disorder. These variations of the diagnosis' context and style represent what Bakhtin (1986) called a speech genre. Although there are as many forms of genres as there are types of communication, each genre has its own style, places its own demands, and commands a specific type of knowledge. Those individuals who are fluent in one genre can be silenced when confronted with another:

> Many people who have an excellent command of a language often feel quite helpless in certain spheres of communication

> precisely because they do not have a practical command of the generic forms used in the given sphere. . . Here it is not a matter of impoverished vocabulary or style, taken abstractly: this is entirely a matter of the inability to command a repertoire of genres of social conversation (Bakhtin, 1986, p. 80).

Importantly, Bakhtin (1986) notes that the inability to converse in a given genre is often the result of discomfort and unfamiliarity, rather than lack of intellect. This appears to be the case in HD misdiagnosis. It is not that the psychiatrists making these misdiagnoses are inept, but rather are framing HD symptoms through a psychiatric, rather than a neurological or neuropsychiatric, diagnostic genre. What these participants, along with the psychiatric case studies, exemplify is more substantial than just an awkward distance between the epistemologies of two disciplines that both happen to study the brain. Rather, these experiences illustrate how these epistemologies are enacted and applied to individuals, and how misdiagnosis is subsequently translated to inaccurate treatment. Therefore, what has thus far been described as a disjuncture, or "great divide", between two disciplines can be stated more accurately to be a diagnostic problem of genres. A critical difference between the latter and the former is that, regarding the latter, Bakhtin provides a means to ground these epistemological problems in the actions of specific individuals by explicitly connecting language to actual utterances (i.e., misdiagnoses). We are thereby directed to the dialogical utterance of the misdiagnosis as the event that actually connects epistemology to everyday interactions.

In this context, it is specifically the utterances elicited within the context of HD misdiagnosis that place psychiatrists into a diagnostic problem of genres. Working within the genre of psychiatric diagnosis, it is unsurprising that an individual presenting with both a familial and biographical history of Major Depressive Disorder is diagnosed with Major Depressive Disorder (e.g., Duesterhus et al., 2004), or that an individual, such as Grace, who presents with anxiety and twitches is diagnosed with Tourette Syndrome rather than HD. The diagnostic problem of genres faced by psychiatrists is further compounded by the inadequate presentation of HD within the DSM-IV. The fact that it is neurologists, rather than psychiatrists, who formally diagnose HD is obviously related to this omission. However, even though psychiatrists are not able to diagnose HD, ample evidence has been presented that they can *mis*diagnose and subsequently mismanage the treatment of individuals with HD (e.g., Tost et al., 2004). The DSM-IV also fails to offer any suggestions or warnings to psychiatrists, through a discussion of differential diagnoses, that they might need to approach individuals seemingly presenting with psychiatric problems from an alternative genre.

It is this omission from the DSM-IV that returns us to the work of Bruno Latour (1987; 2005), specifically his discussion of metrology. Neurology and psychiatry are related and frequently harmonious disciplines, but an ambiguous disease such as HD accentuates their lack of a common language, or shared speech genre. Stated in Latour's terms, these two disciplines lack a system of metrology, which he defines as a system of standards allowing for observations made at one location to be interpreted at another:

> Since without standards like the watt, the Newton, the ohm, the ampere, that is, without the *Systeme International d'Unites*, there would be no global of any sort because no locus would have the 'same' time, the 'same' distance, the 'same' weight. . . All sites would be incommensurable for good (2005, p.228).

When encountering an illness of complex and ambiguous properties, such as HD, the fissures between these two perspectives on the brain become activated. In lacking a metrology, or a system of standards, there is no indication for clinicians encountering a set of ambiguous symptoms on how to proceed, signalling that they might need to convert their observations into those of a related discipline.

The absence of references to HD misdiagnosis in the DSM-IV thus takes on new significance. The DSM-IV, by providing uniform definitions and descriptions of mental disorders, acts as North American psychiatry's metrological artefact. It is precisely within such a text that guidelines should be provided for clinicians for when they might need to use a different system of observation, or diagnostic genre. While the DSM-IV omits this information, the *International Classification of Disease* (ICD-10), published by the World Health Organization (2007) and used by European clinicians, includes an entry of HD as a neurological condition, in addition to its entry as a precursor for dementia. The ICD-10 provides a system of categorization that is more sensitive to the ambiguities between psychiatry and neurology, as it includes diagnostic categories for both physical and psychiatric ailments. However, the ICD-10 also has shortcomings, as the HD entry is particularly sparse and omits a number of key features of the disease, including many of its psychiatric components. Both texts require alterations to adequately address the diagnostic problem of genres highlighted by HD. Given that psychiatrists are relying on a tool that does not include important psychoneurological information, it is perhaps unsurprising that the clinicians attending to Grace, Becky and the numerous other individuals with HD referred to in this manuscript, did not initially suspect HD.

While HD has been used to emphasize these disciplinary ambiguities, many other diseases also straddle the divide between neurology and psychiatry. For instance, Pick's Disease, Korsakoff's Syndrome and Creutzfeldt-Jakob Disease are neurological conditions that feature psychiatric symptoms that are not documented within the DSM-IV. As with HD, the psychiatric symptoms of Creutzfeldt-Jakob Disease often manifest before the neurological symptoms (Spencer, Knight, & Will, 2002), and Pick's Disease has been often mistaken for a psychiatric disorder (McKhann et al., 2001). Perhaps most prominent, however, are the psychiatric effects of multiple concussions, with such injuries garnering significant media attention after being implicated in assaults and suicides perpetrated by several athletes (Schwarz, 2010).

In contrast to these issues, recent social science commentaries have documented psychiatry's increased movement towards neurological methods and explanations. Whitaker (2003) critiqued such decisions in his review of schizophrenia care in America, while Rose (2007) presented the somewhat idealistic accounts of neurologists and psychiatrists who perceived an emanant unity of the two disciplines. While this neurologicalization of psychiatry may be occurring with respect to the descriptions and treatment of some conditions within the DSM, the case of HD presents an important contrast to this process. Specifically, while the rationale for psychiatric pathologies already included in the text may be revised, actual neuropsychiatric conditions remain peripheralized both within the text and the discipline of psychiatry.

Conclusions

Psychiatry has been both externally critiqued from a number of vantage points (e.g., Brown, 1990; Good, 1993; Rosenhan, 1973) and questioned internally, particularly regarding its relationship with neurology (e.g., Price et al., 2000). However, the majority of these commentaries focus on either the epistemological structure of psychiatry or the nuances of clinician–patient interactions. This report has sought to unify these two perspectives by highlighting the epistemological disjuncture between psychiatry and neurology, and its actual consequences for living individuals. While these consequences are suggested in numerous misdiagnosis case studies (e.g., Duesterhus et al., 2004), this report has added nuance to this dialogue by offering the accounts of individuals living with HD. Their experiences underscore the pain, confusion and grief that can

result from misdiagnoses, which are not made particularly salient in case studies or conversations on epistemology.

With an accurate genetic test available for HD, one would presume few reasons for misdiagnosis. However, the benefits of this genetic technology are limited, as an estimated 25% of newly diagnosed individuals have no prior knowledge of HD in their families (Almqvist et al., 2001), giving these individuals few reasons to seek out testing. Additionally, as many as half the individuals with HD avoid disclosing their illness to family members and doctors, due to the stigma associated with the disease, which further complicates genetic testing (e.g., Rawlins, 2010). As this article has highlighted, a further complication exists in that before a genetic test for HD can be performed, a clinician must first consider HD as a possibility. Given the reports on the progression of HD (e.g., Duff et al., 2007), there is a substantial likelihood that this clinician may be a psychiatrist. However, the omission of important information on HD from the DSM-IV, paired with the "great divide" between neurology and psychiatry (Price et al., 2000), situate these clinicians at a disadvantage and accordingly place their patients at risk of misdiagnosis.

This study's participants and the neuropsychiatric features of HD highlight some of the problems with multiple discordant healthcare epistemologies. However, HD also offers a forum and opportunity to make progress on addressing such disjunctures. For instance, although the psychiatric case studies on HD misdiagnosis shared a common mistake, they also shared common recommendations: the need for better understanding between psychiatry and neurology (e.g., Stewart, 1989; Yu, 2004). These statements were echoed in numerous commentaries on the division between psychiatry and neurology, highlighting the need for better education to achieve this understanding (e.g., Price et al., 2000; Schon et al., 2006).

Establishing such an understanding requires more than reforming medical students' education on this topic. A successful remedy also needs to acknowledge that misdiagnosis is the instance where such epistemological divides are operationalized and are translated into very real individual consequences. Accordingly, a pivotal step in decreasing the chances of the misdiagnosis of HD and other conditions would be the implementation of a more comprehensive diagnostic tool. Such a tool would give clinicians an opportunity to translate their observations into the genre of alternative epistemologies and provide an increased sensitization to instances where such translations are necessary. Although a more pluralistic diagnostic guide will not eliminate misdiagnosis, or address many of the other criticisms of psychiatry, it should affect the way its epistemology is enacted, which, as this paper suggests, should have tangible benefits for the individuals encountering it.

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