

THE CULTURAL PARADOX OF PREDICTIVE GENETIC TESTING FOR HUNTINGTON'S DISEASE

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The aim of this article is to perform a cultural analysis of the effects and implications of predictive genetic testing for individuals who have undergone predictive genetic testing for Huntington's disease (HD). Moreover, the analysis aims to relate these effects and the implications of these tests to current initiatives that advocate a large-scale incorporation of genetics and genomics into mainstream health care. The abstract and elusive character of our genes is found to generate a liminal space wherein the affected individuals are situated between normality and abnormality. This juxtaposition of cultural classifications is in turn found to constitute a cultural paradox that might create disagreement in the relations between medical expertise and lay people as genetics and genomics is put to use within mainstream health care.

Keywords: predictive genetic testing, paradox, liminal space, Huntington's disease

I meet up with Jimmy, who is in his early thirties, in his apartment for an interview about his experiences of Huntington's disease (HD)¹, a fatal genetic disease that primarily affects the brain. Jimmy lives in one of the larger cities in the southern part of Sweden. He has a long-term relationship with his girlfriend. His mother has been diagnosed with HD, and as a consequence of the genetic laws that direct the inheritance of HD, the disease and its effects mark the family history on his mother's side of the family. The disease has been traced all the way to his grandmother's mother. Jimmy's mother has now reached that stage of the disease in which the affected individual shows clear signs and symptoms. In order to exclude other possible diagnoses, Jimmy's mother went through a diagnostic genetic test for HD that confirmed that

her signs and symptoms were in fact due to Huntington's disease. Genetic testing is used partly as a diagnostic tool to differentiate between diagnoses, and partly as a predictive tool that gives information on carrier status and individual risk for future disease. As a diagnostic tool, the genetic test is performed in relation to more or less clear and visible signs and symptoms. A clear set of these signs and symptoms is usually not present in relation to predictive genetic testing, since the first appearance of the disease might lie several years, or even decades, in the future. The predictive genetic test for HD provides knowledge on the genetic status of those who choose to go through with taking the test. It does not however offer any knowledge about when those who are found to be gene-carriers for the mutated HD gene

will develop the disease (Kristoffersson 2010: 67–75, 89–98). Apart from confirming that his mother's signs and symptoms were HD, the genetic test and the subsequent diagnosis also put Jimmy at a 50% risk for having inherited the mutant HD gene.² Hitherto, Jimmy's own risk for inheriting the gene had more or less been something that resided in the back of his mind. The confirmation of his mother's HD-diagnosis comprised a transition point (Tibben 2007) that made the potential consequences of the mutant HD-gene obvious for Jimmy:

That's when you'll get it, it's like a punch straight in your face. When you really understand that it's bloody serious. And that I'm in a fix here too...

At this point, Jimmy could choose to wait and see what the future might hold for him. However, he could also choose to undergo a predictive genetic test that would disclose if, or if not, his genome contained the mutant HD-gene. If that were the case, Jimmy would be a genetic carrier or pre-symptomatic patient whose genome contains the mutated gene, even if clear signs and symptoms of HD are not visible at the time when the genetic test is carried out. Due to the scientific development within genetics, the different forms of genetic testing that are available have increased during the last decade and now encompass a range of conditions (Lock & Nguyen 2010: 330–331). In parallel with this increased medico-technological possibility, the prospect for disease prevention has been acknowledged. For example, a discussion article in one of the major Swedish newspapers from 2010 featured three biomedical researchers and experts writing strongly in favour for a wider use of genetic testing within the Swedish health-care system:

A DNA-test would not necessarily make people more worried than other tests within the health-care system. Instead, they can give people an increased knowledge and tools that will enable them to reduce the risk for developing disease. (Björkegren, Nilbert & Syvänen 2010. My translation.)

Similar arguments can be found in the official report from the British Human Genomic Strategy Group on the use of genomics within the British health-care service:

Genomic technologies have the potential to transform the delivery of healthcare in the UK, providing vital insights to support more accurate diagnosis of disease and inform therapeutic decisions – so that more patients get the right treatment at the right time. They can enhance preventive care and enrich our understanding of disease risk, as well as enabling outbreaks of infectious diseases to be controlled faster. (Human Genomics Strategy Group 2012)

These three “snap-shots” convey different viewpoints and different experiences of the ability to make various predictions of future disease in the absence of clear signs and symptoms. From a cultural perspective, these “snap-shots” raise a number of questions. What happens to those individuals who find themselves to be at risk for a genetic disease, and who choose the option of a predictive genetic test in order to receive knowledge about their genetic status? What salient issues can be discerned in conjunction with predictive genetic testing? How can these issues be understood from a cultural perspective? And what implications do these issues have in relation to the use of genetics and genomics as a predictive and preventive tool within mainstream health care?

Consequently, the aim of this article is to perform a cultural analysis, based on ethnographic interviews (see below), investigating the effects and implications of predictive genetic testing on individuals who went through predictive genetic testing for HD. The analysis also aims to relate these effects and implications to current initiatives advocating a large-scale incorporation of genetics and genomics into mainstream health care.

The analysis is guided by concepts obtained from studies in anthropology, ethnology, sociology, as well as science and technology studies. Victor Turner's concept of a liminal space is used as the main an-

alytical perspective. This captures a situation of being in between, as he terms it *betwixt-and-between*, stable and recognized cultural classifications (1979, 1977). Other important analytical perspectives are Åkesson's notion of the gene as an invisible "alien" inside the body (1999), as well as Nikolas Rose's idea of a somatic individuality (2007). Moreover, in relation to the notion of the gene as an "alien inside", the concept of embodied risk or danger also guides the analysis (Lock & Nguyen 2010: 303–329; Kavanagh & Broom 1998). Monica Konrad's notion of a cultural paradox that arises in relation to predictive genetic testing is employed in conjunction with a juxtaposition of normality and abnormality that was observed in the ethnographic interviews (2005: 82). Brian Wynne's investigations of relationships between experts and lay people are used for the section on effects and implications in relation to the utilization of genetics and genomics within mainstream health care. According to Wynne's standpoint, expert notions too often omit emotional and cultural aspects in favour of a more rational and calculative approach on important issues (1996). The concluding remarks of the analysis are built upon this standpoint when they state a necessity for mutual frameworks between experts and lay people in order to make predictive genetic testing sustainable when put to use in mainstream health care.

Regarding the aim mentioned above, HD functions as an illustrative case, as it represents a large group of devastating chronic diseases for which there are currently no cures available. The choice of HD as an illustrative case is a well-established research strategy, since the disease, despite the relative low number of affected individuals, has come to function as a "model disease" within the medical sciences, as well as within the social and cultural sciences. Within the medical sciences, HD shares clinical features with more common brain disorders such as Alzheimer's disease and Parkinson's disease for which there is no clear explanation concerning the causes of these disorders. Within the cultural and social sciences, HD has served as a "model disease" in relation to social and ethical issues, particularly in relation to predictive genetic testing, as HD was

among the first genetic diseases where this form of genetic testing became available within the clinical setting (Tibben 2007; Brouwer-DudokdeWit et al. 2002). The predictive genetic test for HD has been available in the clinical setting since early 1993, following the discovery of the mutated HD gene. In Sweden, an informed consent is required to perform pre-symptomatic or predictive genetic investigations on HD. This means that the provider of the predictive genetic test, which in Sweden is provided by the national health-care system at a genetic clinic, has to be reassured that the individual who is taking the test has been given adequate genetic counselling. The provider must also ensure that the person undergoing the test understands the implication of taking the test. The testing process includes both a consultation with a clinical geneticist, as well as several meetings with a genetic counsellor. The process is designed to provide the individual with enough time to think through his or her decision and it is possible for the individual to withdraw from the testing process at any time. The result is given by a clinical geneticist and followed up at a meeting with a genetic counsellor even if the individual is found not to be a gene-carrier. The individuals undergoing testing and their family members should be offered psychological support suited to their needs. Results are usually followed up for a set period and according to a fixed schedule (Kristoffersson 2010: 95; Socialstyrelsen 2012).

The structure of the article is as follows. The article proceeds with a section on the methods employed in order to obtain the empirical material for this article. The next section will analyse the situation that faced the affected individuals as they discovered that they were at risk of inheriting the HD gene. After this, the consequences of predictive genetic testing upon this situation will be analysed. Finally, the analysis of the ethnographic interviews will be related to the use of genetics and genomics as a predictive and preventive tool in mainstream health care.

Method

A central concern in ethnography is the interpretation of matters that are at stake for particular par-

ticipants in particular situations (Kleinman 1997: 98). In order to understand the way people experience these particular situations, the ethnographic approach often displays a commitment to the particular; this is achieved by using small representative samples instead of larger samples resulting in quantitative generalizations (Smith, Flowers & Larkin 2009: 29–32). This approach will direct the ethnographer to collective (both local and societal) and individual (both public and intimate) levels of analysis of experience-near interests (Kleinman 1997: 98). This strategy within ethnography and ethnology has been employed in a number of studies that investigate different kinds of issues within the context of health (e.g., Alftberg 2012; Browner & Preloran 2010; Lock & Nguyen 2010; Hanson 2007; Konrad 2005; Lundin 1997; Klein 1989).

The ethnographic material for this article was obtained in southern Sweden in 2009–2010 as part of the Basal Ganglia Disorders Linnaean Consortium (Bagadilico) (<http://www.med.lu.se/bagadilico>). Bagadilico is an interdisciplinary research consortium at Lund University focusing on Parkinson's and Huntington's diseases. The study was performed with individuals who in various ways are affected by HD.³ The empirical material mainly consists of in-depth interviews with the participants, whereas observations (which were recorded in the author's field diary, directly after the completion of the interviews) were used as an additional source of knowledge employed in the subsequent interpretation of the interviews. Following Alftberg (2012), it has to be acknowledged that the in-depth interview at times might be seen as a combination of verbal reports and ethnographic observations. The totality of the situation, not only what is verbally reported by the informant, forms the basis for the knowledge produced through the in-depth interview. Here, Alftberg uses the concept "ethnographic interview" as a way to capture this combination of verbally reported material and ethnographic observations (Alftberg 2012: 22). In relation to this article, the ethnographic interview provided an in-depth explorative and detailed knowledge of the experiences reported by the affected individuals. Apart from the knowledge

reported verbally, it also included other aspects that came forward in the interview situation (for example in relation to emotional expressions such as crying at certain points in the interview). The names of the participants have been anonymized in order to protect their privacy. The study was approved by the regional ethical committee at Lund University.

An Invisible "Alien" Inside

Those things and aspects of our existence that, for different reasons and within different contexts, are considered unfamiliar or "alien" have always carried a powerful cultural charge (Åkesson 1999: 121). Arranging different phenomena into cultural classificatory systems is a well-known strategy to create order and control in a world that otherwise would stand out as chaotic and incomprehensible (Åkesson 1991: 57; Lundin & Åkesson 2000: 11). Within the cultural and social sciences, the importance and function played by different classificatory systems is a thoroughly investigated topic (e.g., Foucault 2002; Bowker & Star 2000; Frykman 1993; Åkesson 1991; Douglas 1966).

Within medicine, albeit with a great cultural diversification, classificatory systems are employed in order to diagnose illness and normalize health (Kleinman 1997: 22). These medical classifications split up the world of signs and symptoms into useful categories and models; this became crucial in conjunction with the growth of the modern state (Bowker & Star 2000: 101, 111). For example, the origin of the International Classification of Diseases (ICD) can be traced to the development of the welfare state and its concern with large-scale public health measures and programmes (Bowker & Star 2000: 111, 139–140). And, as noted by Michel Foucault and numerous others, these classifications can also be seen as entwined with the exercise of power within modernity (Hacking 2002: 99–114; Foucault 2000; Lupton 1995; Nelkin & Tancredi 1994).

Traditionally, these "alien features" of our existence were located externally, through such obvious visible criteria as skin colour or sex, which made them easy to incorporate within cultural categorizations. In conjunction with the context of disease,

this aspect can be exemplified by breast cancer. During the eighteenth and nineteenth centuries predisposing causes of breast cancer were traditionally understood in terms of outside influences, like injuries, childbearing, ethnicity or belonging to a certain age group (Schlich 2004: 212). Yet, during the twentieth century, heredity factors became increasingly important as a predisposing factor in relation to disease, often with reference to the developing field of genetics (e.g., Fox-Keller 2010; Nelkin & Linde 2004; Petersen & Bunton 2002). Disease hazards and predispositions, these “alien features” of our existence, are relocated from various outside influences to the internal aspects of our body; this represents a process wherein these dangers and hazards become embodied (Lock & Nguyen 2010: 303–329; Kavanagh & Broom 1998). Epidemiologist Anne Kavanagh and sociologist Dorothy Broom consider that these embodied predispositions, these internal “aliens inside”, are different compared with external hazards and predispositions because “they impose their threat from within – a person both has and is a body” (Kavanagh & Broom 1998: 442).

Today, genetics and genomics are to a large extent centred on the molecular constitution of the human body; DNA, RNA, proteins and various types of other molecules interact with each other and build up the body. New knowledge on our molecular constitution is seen as having profound implications, since it offers a greater understanding of the finest details of disease processes. Moreover, the impact of this development might also be seen within the context of diagnosis and prevention of disease, as it offers a more precise measurement of disease processes and even a reconceptualization of disease classifications (Shostak 2010: 251–254; Rose 2007: 13). According to Nikolas Rose, this development has the potential to reorganize the relations between individuals and biomedical expertise through a reshaping of the way in which human beings relate to themselves. Increasingly, we see and describe ourselves as “somatic individuals”, as beings “whose individuality is, in part at least, grounded within our fleshy, corporeal existence, and who experience, articulate, judge, and act upon ourselves in part in

the language of biomedicine” (2007: 6, 25–26). However, the anthropologist Monica Konrad points out that this picture might be too simplistic, as individuals are not “simply passive recipients of information given to them by clinical professionals” (2005: 63). The ethnographic interviews made within this study involving the experiences among the participants of being at risk, indicated that the participants evaded the idea of the statistical 50% ratio, which only gives a general depiction of the pattern of inheritance concerning this embodied danger that the participants faced. Instead of this general statistical notion, the participants tried to make sense of this potential and abstract “alien inside” by invoking various perceivable aspects within their everyday life.

Patricia is in her thirties and lives in a medium-sized town in the south of Sweden. She has no children and at the time I met her, she lived on her own. Patricia’s situation resembles Jimmy’s, as she came to understand the significance of HD and what it meant for her own future when Peter, her father, was diagnosed with the disease, who in turn inherited the mutated gene from his mother. At the point of her father’s HD diagnosis, Patricia found herself at risk for having inherited the mutant HD gene and she came to the conclusion that she wanted to go through predictive genetic testing in order to find out if she carried the mutant HD gene. However, before she did the test, she tried to make sense of the circumstance of her being at risk by thinking in a way that was tangible for her:

Yeah, you know... when you start adding up on things, you can’t really put your finger on it, but if you ransack yourself, if you really look at the whole picture, you do feel, in some sort of way, that: Yes, that’s the way it is. Of course, I have it. And then I told my mother and the rest of the family: Yes, I have it. Because I did get poisonous goitre from my grandmother, and I... if something strikes... something strange, then it strikes me.

At this point in time, Patricia did not have any knowledge of whether she in fact was a gene-carrier for the mutated HD gene, but despite this ambiguo-

ity of her genetic status, Patricia pinpointed herself as a gene-carrier for the HD gene. She came to this conclusion on the basis of a perceived resemblance (the poisonous goitre) with her grandmother who had already developed HD. Jimmy's reaction on his mother's HD diagnosis provides an additional illustration:

I was completely convinced that I was carrying the gene. So, I went around and checked, planned what I wanted to do during my last ten years alive. What I wanted to do before I was afflicted, that is. Whatever you do, you'll notice a symptom; you spill something, you drop something. Yes, it's symptoms all the time.

As a consequence of his mother's diagnosis, perceived signs and symptoms of the mutant HD gene mentally dominated Jimmy's everyday life. This aspect was also illustrated by a remark made by Carla, who is in her late fifties and is affected by HD through her husband. At the time of the study, he had recently passed away due to the disease. Carla has two children and they both decided to go through with the predictive testing to resolve their genetic status. They were both found to be non-carriers, but before they were tested, their everyday life was also very much dominated by the prospect of being a gene-carrier: As Carla puts it: "Before they were tested? Oh, yes! Every time something happened... oh, now I've got it."

When the participants learnt that HD ran in their family, their everyday life became filled with perceived signs and symptoms of HD; seemingly random accidents, like dropping something, were taken as palpable signs of the mutant HD gene. Likewise, perceived resemblances, like a shared history of coming down with the same diseases, were also taken as palpable signs of an abnormal genetic status (cf. Shostak, Zarhin & Ottman 2011; Konrad 2005: 61–86). At this point, no knowledge of the participant's genetic status existed, apart from the 50% ratio that constitutes a general and statistical description of the inheritance pattern of the HD gene. Nevertheless, from a cultural perspective, the experiences of

the participants were that of being situated between the cultural classifications of normality (being out of danger) and being abnormal (being a gene-carrier). It should also be highlighted that these experiences of being situated between normality and abnormality also included an element of time. As shown in the citations above, the participants fluctuated in time between their present status of being at risk but not yet tested, and an eventual future status of actually being afflicted with HD. The participants seemed to move directly from the present into an anticipated future on the basis of perceived signs and symptoms of an abnormal genetic status, which they come across in their everyday life.

These unclear and indistinctive conditions can be understood, as mentioned above, through Victor Turner's term "liminal space". The term conceptualizes an existence betwixt-and-between different stable and recurrent conditions that are culturally recognized (1979: 467, 1977: 36–37). Inspired by folkloristic research on rites of transition and ritual processes, Turner discerns three phases in these rites. The first phase constitutes a separation, when the subjects who go through the ritual process are detached from their old places within the society. The intermediate phase occurs when the subject is betwixt-and-between recognized cultural classifications. In the third phase of re-aggregation, the subject returns to a new place or position within the community or society (1977: 36–37). Turner characterizes the intermediate phase as a liminal space within which ordinary cultural and cognitive classifications do not apply. Subjects who are situated within this liminal space cannot be understood and categorized through clear-cut cultural classification as they, being "betwixt-and-between" are "neither-this-nor-that, here-nor-there, one-thing-not-the-other" (1977: 37).

The ethnographic interviews indicated that the response of the participants could not be seen in terms of a passive reception of the 50% risk of inheritance of the mutant HD gene. Instead, an active and highly emotional response could be seen among the affected individuals that could be understood in terms of a juxtaposition of normality and abnor-

mality that involved a fluctuation between being at risk and being afflicted with HD. In other words, the everyday existence of these individuals can be understood as being betwixt-and-between normality or abnormality. Monica Konrad considers that this juxtaposition of normality and abnormality can be seen in terms of a cultural paradox (2005: 82). Thus, to enter this liminal space means that you are forced to manage this cultural paradox and the emotional responses that arise as a consequence of an existence betwixt-and-between these two recognized cultural classifications. The ethnographic interviews indicated that this management came to rely upon aspects that were perceivable within the everyday life of the affected individuals. As a way to rework the unclassified betwixt-and-between character of this liminal space into something that was manageable and possible to categorize as either normal or abnormal, they ascribed accidental events or resemblances with an HD-affected relative as a sign of the mutant HD gene.

What happened then when individuals went through with predictive genetic testing for HD? What impact did the knowledge of whether the affected individuals did or did not carry the mutated HD gene have upon their situation? Were they still situated within this liminal space?

The Impact of Predictive Genetic Testing

Both Jimmy and Patricia decided to go through with taking a predictive genetic test for HD. Here their paths start to diverge, because Jimmy's test revealed that he did not carry the mutated gene. Patricia however, was found to be a gene-carrier and will eventually develop HD. As a consequence of her test result, she obtained a status as a gene-carrier or a pre-symptomatic patient but, as will be shown below, this status did not alleviate Patricia's uncertainty as she struggled to cope with the result of the predictive test. This was also the case for her mother, Emma, who now had to face the difficult fact that the disease would definitely not stop with Peter, her husband, but would also affect her daughter as well.

For Emma, who is in her late fifties, Peter's diagnosis spelled out a future marked by a possible con-

tinuation of HD in her family, but Peter's diagnosis also involved a relief as the family got an answer explaining Peter's irritability and at times aggressive behaviour:

Now I have the answer, you know. And now I know why certain things happened. Because he was... I did come in for a lot of physical stuff, you know. And it could be about such a thing that I'd cooked the wrong kind of dinner for him.

The psychiatric symptoms of HD include personality changes, irritability and aggressive behaviour, as well as disturbances in a person's state of mind, such as depression. The loss of cognitive functions includes deficits of memory and attention, which progresses to dementia in the later stages of the disease. For the affected individual and the family, the psychiatric and cognitive disturbances more than often constitute the most difficult and distressing features of HD, even though these signs and symptoms may appear as less striking compared with the more visible motoric signs (Ross & Tabrizi 2011; Petersén 2001: 16).

Carla's HD-stricken husband also displayed behaviour that at times was troublesome, which nevertheless struck Carla as being part of his personality up until genetics provided a confirmation of HD:

Yes, when we came to know [about HD] I understood certain things that were present already when we met, like his urge to be in control (---) He wanted to know everything about what I did and things like that. But at that point, I thought that this was part of his personality.

Carolyn, who is of the same age as Carla and Emma, and has children who at the time of our interview had not undergone the predictive genetic test, also reports the same kind of thoughts. Carolyn's husband was diagnosed with HD at quite an old age, and in our interview she also talked of the diagnosis in terms of a disclosure that gave an explanation of past experiences: "With all the answers in my hand, I can see a lot of symptoms going on many, many years

back in time. Today I can connect this to Huntington but at that time I didn't, of course."

The crucial point in the cases above resided in the presence of clear and visible neurological signs, which a diagnostic genetic test confirmed as being HD. Therefore, within the context of diagnostic genetic testing for HD, the affected families were able to incorporate the result of the test into a cultural classification that permitted the affected individuals to review past events as a consequence of a disease and thereby being of an abnormal kind. This, however, did not necessarily seem to be the case for those who went through predictive genetic testing, when clear and visible signs were more or less absent. For Jimmy, predictive testing showed that he did not carry the abnormal HD gene within his genome. However, despite the test result, there still resided a small but lingering uncertainty in Jimmy's mind whether he was carrying the HD gene or not:

No, but it is just that I don't know anything about all this; I was not present during the [analysing] process. I haven't seen all those machines that do the work. I don't really know how... how it works. The only thing I know is that they got my blood. That's all I know.

This was illustrated by the uncertainty reported by Carla's response to the test result of her two children, who were found not to be HD-gene carriers: "And just this; somebody telling you that you will not be afflicted, or that you will come down with it. Ok, but what does that really mean? That I will develop it. Or that I will not get it?"

Due to the abstract and invisible character of our genes, the result of the predictive test could not be attached to something obvious and concrete. This basic condition, which is an intrinsic aspect of predictive genetic testing, made it difficult for the affected individuals to create a stable and coherent understanding of their test result and of their genetic status.

Carla's thoughts are significant in those instances when the test result showed that the genome did contain the mutant HD gene.

The first meeting that I had with Patricia took place at the public library in her hometown, and there was nothing in her appearance that gave away that she was a gene-carrier for HD. For me she seemed to be perfectly healthy, with no trace of anything near HD. However, when we later met for conducting an in-depth interview Patricia told me about the symptoms that she in fact could perceive. These symptoms resembled those she had encountered during her childhood and her adolescence, when she and the rest of the family were faced with Peter's mood swings:

Yes, it's this... exactly. These somewhat unbalanced [mood swings]; I can get really, really angry. But that's it, as I said previously and today I can say the same, I can get really angry but there's people who become furious and can get wild without having HD (...). But there's this extra dimension, it's hard to explain but it's this extra dimension, so to speak.

In this quote, Patricia identified her own mood swings as abnormal. She could perceive a difference, an extra dimension, which set apart the way she reacted and what she saw as a normal type of reaction. However, in relation to the presence or non-presence of symptoms there was also ambivalence in her experiences; this became apparent at a later stage in the interview when Patricia talked about the test result and about things that were actually happening to her. Things that might be clear signs of the disease:

I feel both relieved and afraid. Yeah, I'm relieved because I feel just like anybody else, I feel energetic and... well, no problems at all. But at the same time I'm really scared because I feel and sometimes I can think: Oh, shit there's actually something that is happening with me.

Up to this point in the interview, I noticed how Patricia had been composed when she talked about HD and her difficult situation. I knew from our initial meeting at the public library that she was quite used to talking about the disease, but at this

point the emotions were coming through and she started to cry slowly. Something was happening to her. Despite the fact that she felt healthy, she also sensed that something was actually going on inside her body. Ambiguous bodily signs that she had not been able to grasp up to this point were now marking their presence in her everyday life. Patricia's experiences might also be associated with Jimmy's statement about not being able to really fathom the abstract process whereby his genome was found to be free from the HD gene. The only experience that Jimmy had about this process was his blood sample from which his DNA was extracted. For Patricia, the discovery that her genome did contain the HD gene was similarly a knowledge that had its origins in the same abstract and remote technological process. This remote technological process revealed that she sooner or later would develop a disease with a pathological appearance that was still quite intangible with her appearance of being a healthy individual.

The two quotes showed that Patricia, despite the knowledge she had about her genetic status, still experienced her situation as being betwixt-and-between normality and abnormality. A similar observation is made by Konrad who means that "the classificatory line between the categories of the pre-symptomatic and the symptomatic resists unambiguous differentiation as separate diagnostic entities" (2005: 81). This kind of collapse of classificatory lines is also observed in conjunction with other medical conditions, where Forss, Tishelman, Widmark, and Sachs report that women who are notified of having cellular abnormalities when screened for cervical cancer are projected into a "liminal state" where "neither health nor disease was confirmed or excluded" (2004: 307). Undergoing predictive genetic testing offered no direct possibility for the affected individuals to clarify themselves as normal or abnormal with respect to HD. Instead of a clarification of their classificatory status, it seemed that both carriers and non-carriers stayed within the liminal space despite the knowledge offered by the predictive genetic test.

To be exposed to this liminal space with its cultural paradox may not only have caused an emo-

tional effect on those who are situated within the liminal space. It may also pose a radical challenge to the Western biomedical notion of the patient as someone who demonstrates a clear and unambiguous cluster of detectable signs and symptoms (Konrad 2005: 82; Sachs 1995: 504). This later aspect came forward in the ethnographic interviews, and it did so mainly in relation to the diagnosis of HD. Currently, the formal HD diagnosis is made when clear motor symptoms appear, a circumstance that Patricia objected against:

Ok, now they're calling me a gene-carrier. But who is saying that? And who has the right to say that, you know? Saying that from now on you're unwell. From now on, although up to that day when somebody says that you're afflicted you're only a gene-carrier. It kind of feels like that I'm not included.

Despite the situation of being betwixt-and-between normality and abnormality, Patricia still made a claim for the power over those classifications that ultimately define her as afflicted or not afflicted with HD. According to Patricia, this power was not to be held by the medical expertise alone, but should also be granted to the affected individuals. Patricia's reaction illustrates how this cultural paradox not only has an impact on the everyday life of the affected individuals, but how it also contains a struggle over definitions and interpretations between those who are situated within this liminal space and various experts.

In the previous sections, I have shown how the abstract and elusive character of our genes give rise to a liminal space within which the affected individuals are situated as a consequence of being at risk for developing HD. To be situated within this liminal space give rise to intuitions, emotions and actions among the affected individuals that include fluctuation between the present and an anticipated future in their everyday life. From a cultural perspective, these responses and actions on behalf of the affected individuals are seen as a result of a cultural paradox that arise from a juxtaposition of normality and

abnormality within this liminal space. Undergoing predictive genetic testing does not appear to resolve this situation. Despite the knowledge of their genetic status, revealed through the predictive genetic test, the affected individuals are still situated within this liminal space with its juxtaposition of normality and abnormality. As a consequence of this juxtaposition, the boundaries that separate the categories of being at risk for HD, being a pre-symptomatic gene carrier or being symptomatic and afflicted with HD are dissolved in the everyday life of the affected individuals.

As the possibilities of making various disease predictions on the basis of genetic and genomic knowledge expand, the issues that have been investigated above might apply to a far greater number of individuals than those who are affected by HD. In relation to this expansion, the proclaimed right on behalf of those who are situated within this liminal space to define the boundaries of normality and abnormality raises questions regarding the relationship between lay people, medical expertise and the health-care system.

Genetic Preventions in Mainstream Health Care

Currently, genetics is partly transformed from being a diagnostic tool, used in the presence of clear and visible symptoms, to a predictive and preventive tool. When this predictive tool is implemented in mainstream health care, the cultural paradox that was accounted for in previous sections can become relevant in relation to other diseases as well. Preventing a disease might of course imply finding a cure, but today prevention has also taken on another meaning, which according to Nikolas Rose aims at making the future “the subject of calculation and the object of remedial intervention” (2007: 19). A central point in this undertaking is to clarify the consequences of a harmful lifestyle, as well as to provide behavioural choices for people so that individuals can make informed decisions on matters of health and health-related behaviour. Here, the growing understanding of genetics and genomics is seen as a significant contribution (Human Ge-

nomics Strategy Group 2012: 34). The two citations given in the introduction showed how the prospect of genetic prediction is framed by medical expertise in terms of offering the individual citizen access to a powerful source of information in order to maintain or restore health. This desire for a future, open for calculation and medical intervention, can also be seen in economic terms, as the state tries to release itself from some of the responsibilities of the consequences of illness and accident that it acquired during the twentieth century (Rose 2007: 19, 63). In this cultural and political setting, every citizen is required to take an active role for securing his or her well-being (2007: 63).

An increased knowledge about the role of genetics and genomics in relation to disease offers great potential for alleviating suffering in various conditions. For a number of these diseases, such as cancer, preventive efforts and early detection is crucial in order for medicine to be applied to save lives. However, a realization of this potential depends, as noted by the Human Genomics Strategy Group, on “public trust in the application of technologies in diagnosis and treatment” (Human Genomics Strategy Group 2012: 79). An important measure in order to safeguard public engagement and trust in the use of genetics and genomics is education of the public on issues relating to genetics and genomics (Human Genomics Strategy Group 2012: 79). However, the relationship between scientific expertise and lay people also depends upon the acknowledgement of interpretative differences between the two groups. Sociologist and STS-scholar Brian Wynne makes the critical remark that expertise of all sorts often “tacitly and furtively impose prescriptive models of the human and the social upon lay people” and that these prescriptive models are “implicitly found wanting in human terms” (1996: 57). The critique of Wynne is directed towards conceptions of lay people in which their response towards expertise is seen in terms of a rational-calculative model. The main critique made by Wynne towards this model concerns the way it put too much emphasis on cognitive dimensions at the expense of the cultural dimensions of public response to expertise (Durant 2008: 7). In-

stead, all prescriptive models on the relationship between lay people and expertise have to acknowledge the “need to recognize hermeneutical differences” (Wynne 2008: 22) which might arise and even diversify the relationship between scientific expertise and lay people.

Predictive genetic testing for HD harbours, as shown by the ethnographic interviews, a cultural paradox in relation to classifications where the invisible “alien” inside give rise to intuitions, emotions and actions that are quite far removed from those rational and calculative approaches that are envisioned in the citations in the introduction. The ethnographic interviews reported in this article also showed that knowledge about whether the participants did or did not carry the mutated HD gene did not totally resolve these hermeneutical differences. Even though the knowledge provided by the predictive test resolved the issue of the participants’ genetic status, there still seemed to exist a hermeneutical difference in relation to the question of normality and abnormality. This difference points towards the challenges that reside in conjunction with a large-scale implementation of genetic tests in the health-care system, since notions of genetic risks and genetic status seldom contain neutral aspects. This absence of neutrality continued to mark its presence in the empirical material on HD, despite the fact that the predictive genetic test itself provided a clear-cut answer about the genetic status of the participants. Furthermore, these challenges concern not only those who are directly involved, patients and medical expertise, but also those social and cultural scientists who study the scientific development within genetics and genomics from various disciplinary viewpoints.

In relation to these differences between lay people and scientific expertise, the importance of an establishment of a mutual framework becomes a crucial measure in order to avoid a diversification and alienation on behalf of those affected. These mutual frameworks have to acknowledge the multiplicity of meanings, viewpoints and practices that exists in relation to predictive genetic testing, as well as the informational requirements generated by this multi-

plicity (Bowker & Star 2000: 297). The establishment of such a common framework between lay people and scientific experts would make predictive genetic testing and disease prevention sustainable if it is to become part of future mainstream health care.

Notes

- 1 Huntington’s disease is caused by a mutation in the HD gene and the pattern of inheritance is autosomal dominant, which means that a child of an affected parent has a 50% risk of inheriting the mutated gene (Huntington’s Disease Collaborative Research Group 1993). In Sweden, the prevalence of HD is about 1/17,000 individuals (Kristoffersson 2010: 94), which means that there is about 1,000 individuals who are diagnosed and afflicted with HD. The disease is characterized by a combination of neurological, psychiatric and cognitive symptoms. In general, the onset of the neurological symptoms appear at an age of between 35 to 45 years, and the progression of the disease always leads to death within 15–20 years after the onset of the neurological symptoms (Ross & Tabrizi 2011). The formal clinical diagnosis is based on the presence of unequivocal signs of motor dysfunction (Huntington Study Group 1996). These neurological symptoms include disturbances in the movements of the afflicted person, mainly causing involuntary movements. The psychiatric symptoms of HD, which most often are present before the onset of the neurological symptoms, include personality changes, irritability and aggressive behaviour, as well as depression (Johnson et al. 2007; Julien et al. 2007). HD also includes cognitive disturbances including deficits in attention that progress to dementia in the later stages of the disease (Stout 2010).
- 2 The concept of risk has been crucial in relation to genetic diseases; it has been investigated from a wide range of disciplinary approaches (e.g., Shostak, Zarhin & Ottman 2011; Lock & Nguyen 2010; Etchegary 2009; Hallowell et al. 2004; Cox & McKellin 1999; Kessler & Bloch 1989).
- 3 Eligible participants in the study were those who in various ways have come in close contact with HD. Participants were recruited through advertisements on the Internet, as well as through personal visits made by the author to various meetings for HD affected individuals held by patient organizations. HD is a sensitive and difficult topic for many of those who are affected. Consequently, the decision was taken to let the number of participants included in the study be based only upon those individuals who the author came in contact with as a result of these advertisements and personal visits. These individuals were then sent additional information (including the form for informed consent) and

were also given time to further reflect on their participation and on the informed consent. Semi-structured interviews were performed with eleven participants at a place of their choice, and the interviews lasted between 1.5 and 2.5 hours. An interview guide was employed for the interviews, but used in such flexible way that the interviewees could go into various aspects regarding their experiences of HD on their own terms. All interviews were recorded by the use of a digital voice recorder, and all interviews were subsequently transcribed *in verbatim*, and each transcript was then read and re-read until a number of themes were identified in the transcript. All citations from the interviews used in this article have not been altered into grammatically correct English, but kept in their original format.

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