Examining Predictive Genetic Testing Using Huntington’s Disease as a Model

A predictive genetic test uses an individual’s DNA to foretell the development of a disease prior to the appearance of symptoms. B.J. Henderson et al.’s article “How People Make Decisions about Predictive Genetic Testing” in the *Journal of Psychology and Health* reports that recent years have brought huge developments in this field and that more can be expected in the future as scientists have started to learn how certain genes factor into the expression of common disorders, such as cardiovascular disease and diabetes (Henderson et al. 514). Moreover, Henderson et al. underscore that “An inevitable consequence of these findings is that more individuals will be faced with the decision of whether to take a predictive genetic test than has previously been the case” (514).

However, despite the increasing likelihood of individuals and families facing this decision, the conversation on predictive testing appears to be largely ethical and focused on if it is right or wrong to take a predictive genetic test for a fatal disease with no treatments or cures available. For example, in an article in the *Journal of Medical Ethics*, author Gwen Terrenoire asks, “What ethical justification can be found for informing a person that he or she will later develop a lethal disease for which no therapy is available?” (Terrenoire 79). Similarly, *Huffington Post* author and Research Program Coordinator at the Johns Hopkins Berman Institute of Bioethics Nathan Risinger contends that individuals simply do not want to be tested for a fatal disease with no known cure, and, due to the lack of a cure or treatment, knowing the results of the test “doesn't -- in this case – help [them] ”(Risinger). Discussions such as these, however, may fail to take into consideration that the decision-making process for this test is
unique for each individual and may not in all cases be covered by blanket statements such as defining a decision in terms of right or wrong or proclaiming the results of a test are useless without a cure available.

Thus, when the conversation is fixed on such questions, it may become easy for people to make snap judgments that they should not test for a fatal, adult-onset, progressive disease. Risinger’s article implies that individuals would only want to discover their genetic predisposition for a disease in order to be cured, and otherwise they would not find the test to be of any service. However, as will be discussed, there may be other, more pragmatic reasons individuals choose to test that make the predictive genetic testing useful to the at-risk person in other spheres. Studying how the test results can serve as a practical tool may give better insight into the motivations of those who are appalled by the idea of taking a predictive test for an untreatable fatal disease.

It may be valuable, then, for us to explore an existing predictive genetic test as a model to gain a deeper understanding of the myriad angles and arguments surrounding such a decision. Using this approach, I will investigate the predictive genetic test for Huntington’s Disease, analyzing the factors that influence patients to take the test, discussing the ramifications of possible outcomes, and addressing the overall impacts of the predictive test. Exploring the factors that influence an individual’s decision when confronted with the option to test, rather than merely negotiating the ethics of the issue, might give a better picture as to why some choose to take a test with a possible devastating outcome. Instead of viewing predictive testing as simply a matter of right and wrong, we might expand our thinking to see the decision depends more on individual choices and circumstances.
I have chosen Huntington’s Disease (HD), in part, as a model for this discussion because it is an autosomal dominant fatal genetic disease. Therefore, if a parent has the disease, the child has a 50% chance of inheriting it (Taylor 137-149). The disease is caused by a repetition of the sequence of nucleotides CAG in a certain place in the person’s genome. HD manifests in a wide variety of kinetic, cognitive, and psychiatric symptoms. The symptoms usually begin in a patient’s thirties to forties, but they can also begin later in life. Death typically occurs 15-20 years after the onset of symptoms. Currently, there is no treatment or cure available. The certain terminality of the disease makes the predictive test controversial (Taylor 140).

Currently, recommendations do exist to guide clinical practice of the HD predictive genetic test. The guidelines were written in 1994 by the World Federation of Neurology and the International Huntington Association, but were revised and updated in 2012 (MacLeod et. al 222). The guidelines are organized in terms of access to the test, what information must be communicated, delivery of results, and post-test counseling. The aim of these guidelines is to make the test fair and comprehensible. For example, section COM 5.3.1 states that extra counseling should be provided to patients having difficulty coping with the results of their test (226). These standards are not rigid laws but recommendations that have influenced predictive testing practice in other late onset diseases.

The HD predictive genetic test, developed in 1993, seeks to find out if the abnormal gene for HD is present in the individual’s genome (Taylor 138). If an individual tests gene-positive, it is certain that he or she will develop Huntington’s disease, and his or her children will each have a 50% chance of developing the disease. Testing gene-negative indicates that the individual will not have the disease. One can imagine, then, that taking a predictive genetic test for a fatal disease with no treatment can be an incredibly difficult decision to make. A study by Sandra D.
Taylor explored the “test or not to test” dilemma. Sixteen at-risk individuals who had already decided whether or not to take the test were interviewed. The study found that all participants viewed the test as a way to acquire “significant life knowledge by accurately refining a 50% risk for HD to either zero (a gene-negative result) or to 100% (a gene-positive result)” (Taylor 140). The choice not to test is respected by all participants in Taylor’s study, even those who chose to test. Interviewees also take into consideration self-assessments, such as coping abilities, emotional stability, and their current life stage.

Participants opting to test in Taylor’s study focused on moral responsibilities towards others, such as spouses and children. This idea is echoed in J.A. Smith et. al’s article “Doing the Right Thing for One’s Children” in the Journal of Clinical Genetics. This article summarizes the findings from a study that Jonathan Smith, M. Stephenson, C. Jacobs, and O. Quarrell conducted with nine participants who were asked whether they would agree to genetic testing for Huntington’s Disease and why. Smith and others point out that the participants’ decisions and explanations varied depending on the participants’ ages, number of children, and outlook on their own and other family members’ health. Some participants in the study chose to take the test to help their adult children with reproductive decisions. For example, Angela has an adult son and daughter who do not have their own children yet. She is taking the test because she feels it is her duty to give her children the important information about her own genetic predispositions so they can make their own responsible reproductive decisions, for example, in deciding the number of children they might choose to have (Smith et al. 419).

Choosing to test is a difficult decision because receiving either result can be accompanied with complex benefits and/or adverse consequences. Receiving a gene-positive result can have staggering affects on a person; the person now knows he or she will experience a terrible, fatal,
neurodegenerative disease. While this news will be extremely difficult to adjust to, it can ease the anxiety of not knowing and assist in planning for the future in matters such as reproductive decisions. Testing gene-negative has the obvious reward that a once “at-risk” person can now abandon such a label and live without worry of having HD. However, Taylor argues that there can even be negative consequences upon receiving this heavily favored result, explaining that individuals may experience “a likely need for readjustment of self-identity from being an ‘at risk’ person” (Taylor 138). If one is accustomed to being from a family with HD and they now find out they do not have it, there may be a change in family roles and feelings of guilt. Individuals who have family members suffering through HD may feel intense self-reproach that they escaped the disease. This illustrates that both outcomes of the taking the test can possess benefits as well as drawbacks.

Those who chose not to test in Taylor’s study did so for a number of reasons, such as their ability or inability to handle a difficult change in situation, traumatic family history, or their desire to confront the future (Taylor 143). Some individuals believe that receiving a positive result could debilitate them tremendously and ruin their lives. For example, participant Carol, who is undecided about predictive testing, stated, “I’ll sit here and think… Should I go get tested or what… but then I don’t know what I’d do if I got a positive result… Whether I’d worry myself to death and have a heart attack or what…” (Taylor 146). The knowledge of the results cannot be unlearned, and the potential outcome could present a huge emotional burden. For those who lack strong coping abilities, taking the test could result in serious repercussions. Other justifications for refusing to test stemmed from a familiarity with the disease because of a family member who has suffered from it and from an individual being philosophically opposed to finding out anything about his or her future ahead of time. Steven, a 50-year-old non-testing
participant, claims, “I’m a person who thinks life would be very dull if you had a crystal ball… I’d sooner not know what’s going to come up in the future” (Taylor 143). The disparity of decisions with regards to testing and the myriad of reasons in favor of or in opposition to predictive genetic testing helps to illustrate why a simple debate of ethics may not yield a complete understanding of the complexities of deciding to test for HD.

Simply because a certain technology is available does not always mean it should be implemented. As citizens of the 21st century, we are constantly encountering new technology, in the medical field especially, and we are constantly wrangling with the ethics of such technologies. Predictive genetic testing is an issue that may become vital in many aspects of our society, such as policy, insurance, and medicine, and if we continue to think about these issues in binary terms, we run the risk of making enormously uninformed decisions. An approach to genetic testing that takes into consideration all of the possible impacts, not just on the patients themselves but on current and future family members, will present the greatest probability of success, whether that refers to a patient coping with a condition or a doctor figuring out an approach to tackling it.
Works Cited


Smith, JA, M. Stevenson, C. Jacobs, and O. Quarell. "Doing the Right Thing for One’s Children: Deciding Whether to Take the Genetic Test for Huntington’s Disease as a
