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Disclosure of At-Risk Status for Huntington's Disease: Is There a "Right Time?"

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DISCLOSURE OF AT-RISK STATUS FOR HUNTINGTON'S DISEASE: IS THERE A "RIGHT TIME?"

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Introduction

In 1872, American physician George Huntington described a disease that can be traced back to the Middle Ages, when it was referred to as chorea. Chorea is a term that describes the uncontrollable movements that are seen in individuals who are affected by the disease. This disease was found to be hereditary and progressive. Today this disease is called Huntington's disease (HD). In addition to chorea, HD also causes behavioral changes, memory loss, and mood swings. Currently more than 15,000 Americans are living with HD, with at least an additional 150,000 others who have a 50% chance of developing the disease. This disease is passed from generation to generation, destroying families who have to watch their loved ones lose the ability to feel, think, and move (National Institutes of Health, 2009).

HD's ability to destroy entire families stems from its genetic basis. HD is an autosomal dominant genetic disease. This means that the children of those suffering from HD have a 50% chance of developing the disease. If the genetic basis for this disease is inherited, the disease will manifest. Those at risk for HD have the choice of being tested for the genetic marker. The test can definitively tell an individual whether he or she will develop the disease. However, knowing if one will develop the disease cannot stop the disease from developing. Currently there is no cure or treatments to slow the progression of HD (National Institutes of Health, 2009).

The choice to know or not to know if one will develop HD becomes part of every day for the individuals at risk, their spouses, their children, their employers and their insurers. The decision on how to disclose the risk for HD to others is a constant source of conflict and turmoil, especially within families. Families with a history of HD must decide who is allowed to know, when they should be told, what they should be told, and who should do the telling (Quaid et al., 2008). These decisions can wreak havoc on a family and can have unintended effects.

Problem/Purpose

The disclosure of at-risk status for HD can be a complicated and stress inducing process. The most difficult people to disclose this information to are most often those who are also at-risk, notably the children of individuals who are themselves at-risk for developing the disease or know that they will develop the disease. How and when a child finds out that he or she is at risk for a debilitating disease can have a tremendous impact on the development of the child. The purpose of this thesis is to assess the ideal course of action in dealing with how and when to tell a child that they are at risk for HD.

Research Methods

This research was conducted using the body of information found in available journal sources. The research that had been done previously on the topic was analyzed and synthesized to identify common findings or patterns. Since much of the relevant

research had been done using qualitative methods, a unique synthesis of statistical data could not be performed. Instead the findings were compared and new commonalities were found. These commonalities were then used to make suggestions concerning disclosure of at-risk status for Huntington's disease to children.

Definitions

One of the main concepts in this research is being "at-risk." The term "at-risk" is operationally defined as having a parent or grandparent develop Huntington's disease. Every child of a person with HD has a 50% chance of developing HD. If a child's grandparent developed HD, the child's parent would have a 50% chance of developing HD and the child would have a 25% chance of developing the disease. The term "disclosure of at-risk status" is operationally defined as the time when one individual informs another individual of his or her risk for developing a genetic disease.

Literature Review

Biochemistry Background of Huntington's Disease

As stated above, HD is a progressive disease that causes the loss of control over movement, behavioral changes, and memory loss over time. The onset of HD is generally slow and insidious, making it hard to distinguish when an individual transitions from being a risk for HD to having HD (Maio et al., 1992). On average, symptoms of HD begin around 40 years of age (Meiser & Dunn, 2000), although the disease has been seen in children as

young as 2, and adults as old as 70 (Quaid et al., 2008). After the onset, individuals with HD generally live for 10-15 years. Currently there is no way to cure the disease or to slow the progression of the disease (Meiser & Dunn, 2000). Patients suffering from HD commonly die from infection (such as pneumonia), injuries related to a fall, or other complications from HD (National Institutes of Health, 2009).

The major signs of onset of HD are the start of mood swings as an individual becomes increasingly and uncharacteristically irritable, apathetic, passive, depressed or angry (National Institutes of Health, 2009). Some patients affected by HD may start with the onset of chorea, or uncontrollable movements. The uncontrollable movements begin in the fingers, feet, face, or trunk. HD also affects patients' judgment, memory, and other cognitive functions. For example, individuals with HD can have trouble driving, learning new things, remembering a fact or making a decision. As the disease progresses, concentration on intellectual tasks becomes increasingly difficult. The disease can reach a point where speech becomes slurred and vital functions decline. Some individuals lose the ability to recognize family members, but most remain aware of their environment and are able to express emotions (National Institutes of Health, 2009). Stern and Eldridge (1975) found that those who have HD, or are at risk for HD find the physical disturbances to be the most troubling feature of the disease. Meanwhile, relatives, including spouses, of individuals with HD found the mental disturbances to be the most troubling feature of the disease if their relative or spouse had been affected for less than five years. However, if the

disease had progressed past the five-year mark, physical disturbances were found to be the most distressing feature (Stern & Eldridge, 1975).

The symptoms of the disease are caused by a degeneration of neurons, as well as from dysfunction of affected neurons before they die. The protein product of HD, huntingtin, forms aggregates within neurons, generally inside the nucleus, which causes the dysfunction and death of neurons (Ross et al., 1999). Huntingtin is a large protein comprised of 3144 amino acids. Beginning at the eighteenth amino acid there is a poly gulatmine tract. This tract generally contains 11-34 glutamine residues in unaffected individuals. In individuals affected by HD this tract contains more than 37 glutamines. The function of normal huntingtin is still undetermined, but with an expanded glutamine tract there is a change in properties. This change in properties is what allows for the huntingtin to aggregate and cause neuronal death (Li & Li, 2004). The neurons that are most affected by huntingtin are in the basal ganglia, which are responsible for body movement and coordination, and the cerebral cortex, which is responsible for perception and memory.

The expansion of the glutamine tract is coded by a trinucleotide expansion on chromosome 4. This trinucleotide expansion is the genetic basis for HD. Individuals who have between 5-35 CAG repeats will not develop HD, but individuals who have 36 or more CAG repeats will develop HD. The CAG trinucleotide expansion was shown to be a highly sensitive and specific marker for the inheritance of the disease by Kremer et al. (1995). In a study 1007 patients who had been clinically diagnosed with HD were tested for the CAG expanded repeat. The patients were of different nationalities and countries of origin. Of

the 1007 patients included, 995 had an expanded number of CAG repeats between 36 and 121, with a median of 44 (Kremer et al., 1995). The CAG expansion is what is used to test for the presence of HD in predictive genetic tests.

Genetic Testing

Unlike other predictive genetic tests, an expanded CAG repeat corresponds to a nearly 100% chance of showing the symptoms of the disease (Quaid et al., 2008). Also the number of repeats has been found to correlate inversely with the age of onset of the disease (Snell et al., 1993). It was found that the more repeats present the earlier the onset of the disease will occur. It is also likely that a larger number of repeats leads to an earlier progression and more severe symptoms (Snell et al., 1993). As a result, in an individual who is tested for HD not only finds out whether he or she has the mutation, but also when the disease is most likely to develop and how severe the symptoms will be.

Significant concerns have been raised in the past about whether or not it is ethical to offer a predictive test when there is no known treatment to delay onset or to slow the progression of the disease. The concern was that the results of the predictive testing would facilitate additional stress, depression and even suicide for those who found out that they would develop HD (Hayden, Block, & Wiggins, 1995). The potential benefits of having the genetic test include relief from uncertainty, the chance to avoid passing the trait on, and prudent future planning (Codori, Young, Miglioretti, & Brandt, 1997).

The major concern over testing for HD is the prediction that the results would increase suicidal behavior in those who tested positively. It was found that the frequency of suicide was higher in patients with HD and in their relatives, including their spouses, than in the general U.S. population (Maio et al., 1993). A separate study researched the rates of suicide, suicide attempt, and psychiatric hospitalization after a positive HD test (Almqvist, Bloch, Brinkman, Craufurd, & Hayden, 1999). This study found that out of 4,527 participants, five successfully committed suicide, 21 attempted suicide and 18 were hospitalized. This was less than 1% of the participants. Those who were among that 1% were more likely to have had a psychiatric history less than five years prior to testing and were more likely to have been unemployed (Almqvist et al., 1999). This study seems to suggest that the concerns about increased suicide risk after testing may not apply to all of those who are at risk for HD.

To address the concerns raised about the less severe effects of testing for HD, Hayden et al. (1995) studied the quantitative and qualitative psychosocial aspects for 400 persons at risk for HD before and after being tested. For the quantitative aspects the participants in the study were given the Beck Depression Inventory and The General Well-being Scale before being tested and 7-10 days, 6, 12, 18, and 24 months after receiving their results. For the qualitative aspects the participants were also interviewed at various intervals. Of the 400 participants, 60% received a decreased risk prognosis, meaning it was unlikely that they would develop HD. Compared to their initial scores most of the participants who received this result had much lower levels of psychiatric distress and had

an overall improvement of psychological health at the six and twelve months follow-ups (Hayden et al., 1995).

While most of the participants receiving a decreased risk result had an improvement, there was a sizeable portion of those who received this result (10%) who had an adverse reaction to their result (Hayden et al., 1995). This was found to be because of three main reasons. The first was having previously made major, irreversible life decisions, such as marriage, child bearing, and financial decisions, based on the belief that he or she would develop HD. The second is survivor guilt from having a parent or sibling who developed HD, while the participant was found to be healthy. The third was no longer having part of their identity. Believing that one will someday develop HD can allow individuals to use the disease as a psychological crutch, without which these people do not know how to function. The critical time period for these individuals was between two months and two years after receiving the results. During this time period these individuals are at most risk for maladaptive coping and in need of additional support (Hayden et al., 1995).

The other 40% of participants involved in this study received an increased risk result. These individuals were expected to develop HD at some point in the future (Hayden et al., 1995). Contrary to expectations, these individuals also showed an improvement in overall psychological health at the follow-up sessions. While the improvement in scores was less than those in the decreased risk group, it is important to note that there was still improvement. Knowing the result of the predictive test, even if it indicates that he or she

has the mutation, reduces the anxiety and uncertainty and provides an opportunity to plan for the future (Hayden et al., 1995). It was found that there was an adjustment period after receiving the results when there was some emotional distress, but past this period most people reached acceptance. For these individuals the first year after receiving the results from the genetic testing was the time in which they were most likely to cope ineffectively and need additional support (Hayden et al., 1995).

Codori et al. (1997) showed similar results in a study of 160 at-risk adults. They found that the individuals in both the negative and positive result groups had distress measures below clinical cutoffs even after receiving their results. Those who tested positive for HD had a short period when their scores were below baseline, but over time the scores returned to the original. It was hypothesized that the lack of negative effect was due to self-selection into the study. The participants in the study had to agree to take part in the genetic testing, so those who did not want to know, or would have reacted more negatively, self-selected out of these types of studies (Codori et al., 1997). Additional studies have shown that adjustment to results depends more on psychological adjustment before testing than it does adjustment after testing (Meiser & Dunn, 2000).

Adjustment can be mediated by a variety of factors including marriage status and nearness to age of onset (Codori et al., 1997). As expected, individuals who were closer to the age of onset took longer to adjust to a positive result. However, contrary to what was expected, those who were married took longer to adjust to a positive result than those who were not married (Codori et al., 1997). Also contrary to what was predicted, those who did

not have children had higher levels of hopelessness than those who did have children. It had been expected that those with children would then feel guilt for the possibility that the disease would be passed on. While these individuals may have felt guilt, it did not manifest itself as hopelessness on the scale that was being used in the study (Codori et al., 1997).

Disclosure of At-Risk Status

While these studies have shown that most of the people who participate in genetic testing do not have an overall negative psychological reaction, only 10-20% of those at risk for HD undergo testing (Meiser & Dunn, 2000). A recent study done by Quaid et al. (2008) found that there were two overarching patterns in those who chose not to be tested: careful concealment as an act of self-preservation and preserving hope. Quaid and her colleagues found these two patterns by interviewing at risk individuals who had opted not to take the genetic test. It was found that the risk, the hope, and the uncertainty affect every aspect of a person's life. For some individuals the hope can outweigh the stress of uncertainty. They also found that the choice to know or not to know becomes part of every day for the individuals at risk, their spouses, their children, their employers and their insurers. The decision on how to disclose the risk for HD to others is a constant source of conflict and turmoil, especially within families. The most problematic disclosure for those at risk is how and when to tell their children (Quaid et al., 2008).

While the timing may be difficult, it is recognized by most families that at some point they must tell their relatives about the family's history of HD. Within families there

are many barriers regarding when to tell, what to tell, and who to tell (Forrest et al., 2003). A study looked at these issues within families with late-onset genetic disorders (either HD or hereditary breast and ovarian cancer) by qualitative interviews. The study found that those at risk for HD believe that the responsibility of telling family members of their at-risk status belongs to relatives as opposed to health professionals. The barriers to family disclosure that were found were the pre-existing relationships, patterns of interaction, and tensions and fighting that hinder communication. For example, many family members felt that they should defer to a higher authority within the family to tell other relatives of their genetic risk. Within families there seems to be implicit rules about who gets told, who tells and what to tell. These barriers can then cause more tension and emotional burden on those who know and anxiety and anger from those who find out later in life (Forrest et al., 2003). There is also a generational effect, a belief that that those of the older generation have the obligation to tell the younger generation, but also the responsibility to protect the younger generation. With HD this becomes further confused by the need to know before important life decisions are made.

In addition to the many life decisions that those affected with HD must make while they are still young, young people's experience with HD is important to study on its own because they have different cognitive and emotional capabilities than adults. Understanding HD and the impact it could make on a person's life is important to development and to making life decisions. During adolescence, identity formation occurs. Having a parent that is affected by HD can have a major impact on a child's identity

formation, as will the child's at-risk status (Keenan, Teijlingen, McKee, Miedzybrodzka, & Simpson, 2007). If the child is not aware of HD while forming their identity, they may later have a negative coping experience when they find out. These individuals may experience an identity crisis after learning of their at-risk status for HD.

Families with a history of HD walk a fine line in balancing the need to protect their children for as long as possible, with the need to tell them in time to make informed decisions and to understand the problems that may be to come. This balance becomes more difficult when the child is raised with a parent or grandparent exhibiting signs of HD. Recently a qualitative study was done to investigate the different ways children were informed of their at-risk status and their reaction to the experience (Keenan, Teijlingen, McKee, Miedzybrodzka, & Simpson, 2009). In this study 33 at-risk young people from Scotland were interviewed about their experiences with finding out about HD. These researchers found that different styles of family communication will impact the child's experience of growing up with HD. Some research had suggested that many at-risk children sensed that 'something was wrong' long before they learned about HD, even as young as five. Some of those children realized that whatever was wrong was a secret and learned not to ask questions. These types of reactions can cause unnecessary anxiety (Keenan et al., 2009).

Keenan, et al. (2009) discovered that there are four ways in which children learn about their at-risk status for HD: (1) 'Something is wrong,' where HD is an unknown diagnosis for the family and they search to find out what is wrong with a relative, (2) 'Out

of the blue,' where they find out immediately without wondering what is wrong, (3) 'Knowing but dismissing,' where the child has a vague idea of what HD is but is never exposed to it or worried about their own future, (4) 'Grown up with HD,' where the child learns about HD gradually but HD is never hidden.

From the interviews of young people at-risk for HD, four basic disclosure experiences were found to be prevalent: (1) having always been told, (2) gradually told, (3) HD was kept a secret, and (4) HD was a new diagnosis. Some of the children were active agents in their learning about HD, through listening, watching, asking, and searching for answers. Many of those who grew up always having been told had been exposed to a close relative manifesting symptoms of HD, which may have encouraged the disclosure. Those who were told gradually described a period of initial awareness that was distressing. Those whose parents had kept HD a secret found out about HD in late adolescence or young adulthood. Some of those in this group had a sense that something was wrong but had learned not to ask about the family secret. Those in the group for whom HD was a new diagnosis found the disclosure a complete shock. The individuals in both the 'kept secret' group and 'new diagnosis' group had the least of knowledge about HD. The support needs of these individuals are completely different from those of the other two groups because they were older than the other participants upon learning about HD and had thus made more life decisions without the knowledge of their at-risk status (Keenan et al., 2009).

It was found that those children who grow up with close contact with an affected family member want to know what is wrong and can cope with knowing the truth at an

early age. When these children are not told they feel confused and anxious and can rebel against the protective parenting style adopted by their parents (Keenan et al., 2009). It was found that there is an important distinction between being told of a parent's at-risk status and being told of one's own at-risk status. Reactions to each disclosure can vary and the researchers suggest that the disclosure should be taken at the child's pace. Thus disclosure can be a lengthy process (Keenan et al., 2009).

An earlier study completed by the same researchers found that young people are able to successfully cope with knowing about HD. They found that those children who found out about HD at a young age were more likely to cope successfully than those who found out later on in life (Keenan et al., 2007). In the later study Keenan et al. (2009) found that there was a difference in experience based on whether the child grew up close to an affected family member. Children who did not grow up with close contact coped well by finding out gradually at a slightly older age. Young people whose parents were honest and allowed for questions described their experiences more positively than other young people (Keenan et al., 2009).

Where open communication existed, young people as they matured into adulthood were more cautious about their reproductive decisions (Metcalfe et al., 2008). Children who were allowed to ask questions, seek information on their own timeline subsequent to the initial disclosure, and talk with other family members were able to adjust more quickly. Poor communications of information about the genetic disease led to reproductive choices based on inaccurate information. Poor communication also led to emotionally driven

decision-making in young adults. Without the proper information, young adults were influenced more by feelings of anger and upset than by logic when making important life decisions. Adult children felt that, had more information been provided earlier, some decisions might have been made differently (Metcalfe et al., 2008).

Similar studies on communication of genetic risk have been performed with families at risk for hereditary breast-ovarian cancer. Hereditary breast-ovarian cancer is similar to HD in that it is a genetic disorder that has a late onset and the mutation that causes it can be tested for. However, unlike HD, there is a treatment for breast and ovarian cancer and the mutation that is associated with hereditary breast-ovarian cancer does not cause the disease all of the time. In women, having the BRCA1 mutation, which is one of the two mutations associated with hereditary breast-ovarian cancer (the other is the BRCA2), is associated with a 55-85% life time risk of breast cancer (Tercyak et al., 2001). With HD, those who have the increased trinucleotide expansion of 36 and above will definitely develop HD. While there are significant differences between hereditary breast-ovarian cancer and HD, it may be the closest comparison in terms of disclosure barriers.

A study investigating the communication of BRCA1/2 genetic test results with children found that parents who were more distressed were more likely to communicate their test results to their children (Tercyak et al., 2001). This study was completed using a qualitative interview design with 133 mothers and fathers who were going to be tested for the BRCA1/2 mutation. The participants took a questionnaire to determine their general distress and cancer-specific distress both before and after they were tested for the

mutation. The study found that around one-half of both carriers and non-carriers chose to disclose their results to their children. In families with more than one child, it was most common for the parents to inform all or none of their children, as opposed to telling one child and not the others. The researchers suggested that those parents who disclosed the information from the test might have wanted to have an open style of communication in their family. There was also the result that those individuals who had higher levels of general distress both prior to the test and after the test shared the results with their children more frequently. This finding would suggest that these parents disclose the information to alleviate their own negative feelings (Tercyak et al., 2001).

The presence of children can also affect an individual's participation in genetic testing. Women at risk for hereditary breast cancer are more likely to be tested for the BRCA1/2 mutation if they have daughters (Croyle & Lerman, 1999). Being tested for the hereditary marker for breast cancer is related to the desire to help other family members. Many times individual go through testing so that they are better able to assess other family members' risk, as opposed to their own. It was found that women who shared their test results for the BRCA1/2 mutation with their sisters had a decrease in psychological distress, while those who did not share with their sisters had an increase in distress (Croyle & Lerman, 1999). Studies have found that family influences can affect not only communication, but also risk awareness, genetic testing decisions, and coping strategies.

The disclosure of at-risk status can cause many emotions not only in the child who is learning, but also in the parent who is disclosing. Parents expressed emotions of anxiety,

worry, and concern with informing their children of the genetic disorder. No parent wants to see their children hurt, and informing them of a major family disease will cause pain. The majority of parents reported a lack of support or advice from medical professionals on how to discuss the genetic disorder with children. Knowing what to say and how to say it can be a long, difficult process. Without the support of medical professionals, parents may have taken longer to disclose the information. While many parents felt negative emotions toward the disclosure, none of the parents reported regret about discussing the genetic disorder with their children (Metcalfe et al., 2008).

One of the benefits of informing children while they are still young is that younger children have the opportunity to incorporate their genetic risk into their self-identity (Metcalfe, Coad, Plumridge, Gill, & Farndon, 2008). Older children or young adults would have to reevaluate their self-identity to incorporate their at-risk status. The older children have more concrete aspirations and life expectancies that may have to be changed due to their being at-risk for HD. Being told later can cause resentment and can damage relationships within a family (Metcalfe et al., 2008). In addition to the adjustments a young adult would have to make in their own identity, they are also then closer to the age of onset for their parents.

Due to the debilitating nature of HD, an individual who begins to manifest symptoms will increasingly need a caregiver. Many times caregiving falls onto family members. The necessity of caregiving can also change a young person's life. The most harmful way to disclose at-risk status to a child is to overload (Keenan et al., 2009).

Overloading is where the parent informs the child of his/her own risk, while concurrently informing the child of the parent's need of care. Adjusting to both caring for a parent and learning of one's own risk can lead to ineffective coping. This group of young people has the greatest risk of self-harm and need for support from outside sources (Keenan et al., 2007).

Caregiving to Family Members with HD

Caregiving in itself can be a considerable burden. It can be described as an unexpected career. This is because it generally starts out as part-time assistance that transitions into a full time role. Caregivers witness first hand the deterioration of their loved ones. They also witness the loss of the loved ones' personality while he or she is still physically there, which can be worse than grieving his or her death (Pickett, Altmaier, & Paulsen, 2007). The burden of caring for someone with HD is distinct from caring for someone with other disorders. First, HD symptoms onset occurs earlier than other progressive diseases that include dementia as a symptom; thus, the caregivers of HD are younger than other caregivers. Second, due to the heritability of HD, the caregiver is tasked with seeing first hand what he or she may endure someday. Third, compared to other neurodegenerative diseases, HD has a slower progression, so the caregivers of HD spend more of their lives giving care than those who care for individuals with other diseases. Lastly, individuals suffering from HD are so difficult to manage that many facilities are hesitant to take them in (Pickett et al., 2007). This means that caregivers are rarely able to

seek support from full time care facilities. The prospect of becoming a caregiver can provoke anxiety by itself. It is easy to see how dealing with this along with the new knowledge of one's own risk can be doubly difficult.

To reduce the anxiety and stress of learning about HD and all of the issues that come along with it, including care giving, some individuals seek social support groups outside of their family. This can be helpful in the disclosure process because the young person will no longer feel that they are the only one experiencing the stress and anxiety caused by HD. Support networks can also be helpful in expanding knowledge about HD. However, due to the relative rarity of HD, it may be difficult to form an in-person support group. More recently, online support groups have been made available to individuals dealing with being at-risk for HD (Coulson, Buchanan, & Aubeeluck, 2007).

Support groups provide an additional source of information for children, and allowing them to seek information that is relevant to them. Online support groups, such as the one Coulson et al. (2007) studied, provide both informational and emotional support. Those at risk for HD face unique challenges that other individuals are unable to comprehend, especially the concern over whether to get the genetic test. The online support groups mirror the discussions and support provided by face-to-face support groups. Seeking either online or face-to-face support groups was found to be beneficial in the adjustment process of learning about HD (Coulson et al., 2007).

Significance

Huntington's disease is an incredibly debilitating disease that affects thousands of people in the United States. Every child of a person with HD has a 50% chance of developing the disease, and is referred to as "at-risk." How an individual is told of his or her risk for developing HD can affect many aspects of his or her life. It can affect the types one's hopes and dreams, use of coping mechanisms, decision-making processes, and identity formation. Finding the correct time and the correct words to explain a complex disease can be a difficult process for families. Determining if there is a time period in which children are able to cope best with the disclosure of their at-risk status will aid families in their struggle with informing their children. Also, by determining the most beneficial communication style to use while informing children, families may be able to avoid the trauma and hurt that comes with being informed improperly.

Discussion

Huntington's disease is unique from other diseases because it is both an autosomal dominant genetic disease and a late-onset disease. This provides individuals at risk for the disease with the distinct perspective of having a 50% chance of developing the disease after the age of reproduction (National Institutes of Health, 2009). An individual who is at risk has to worry about his or her own risk, while also worrying about his or her children's risk. Once the symptoms begin to manifest, they will progress until death. The symptoms

of HD - chorea, behavioral changes, cognitive changes, mood swings, and dementia - lead to the individual needing full time care (National Institutes of Health, 2009). The care of a person suffering from HD is generally the responsibility of a family member, most often a spouse or child (Pickett et al., 2007). Those at risk for the disease often have to watch the disease destroy a family member before they themselves develop any symptoms.

Due to all of the issues that are associated with HD, the prospect of developing the disease can cause anxiety, stress, and uncertainty. HD affects individuals' decisions about whether or not to have children, whether they should live in the moment because that is all they have or if they should save their money to help pay for the full-time care they will need (Codori, 1997). Decisions must also be made about whom to tell of their status, when to tell, and how to tell. The most difficult of those is how and when to tell their children (Quaid et al., 2008).

Parents tend to want to protect their children for as long as possible, allowing their children to be children without burdening them with the information about HD. This, however, can lead to children feeling stifled, over-protected, and many times more anxious (Keenan et al., 2009). Parents who actively try to keep the information about HD from their children tend to allow that closed form of communication to expand to all areas of life. This can lead to children feeling a range of negative emotions, as well as rebelling against what they see as an unfair parent. Also, many times children know that something is wrong. This can lead to more stress and anxiety due to the uncertainty. In addition, if a parent waits too long, he or she runs the risk of overburdening his or her child. This occurs

when a parent discloses the information around the same time that he or she is showing symptoms of the disease. The child must then cope with the fear for his or her parent, the burden of caring for the parent, and the knowledge of his or her own risk (Keenan et al., 2007).

Adjusting to the knowledge of potentially having HD can be a painful experience. Learning of one's risk can lead to many issues including suicidal thoughts, testing dilemmas, identity crises, and worry about the future. Being able to successfully cope with being informed of a family history of HD depends on when the person is informed and how the person is informed. Those who are most successful at coping with their at-risk status are those who have 'always known' about HD in some way. By knowing about HD since they were young children these individuals were able to incorporate HD into their self-identity. These children were able to use their knowledge of HD to form their hopes, dreams, and life expectations. Individuals who are told at older ages have to rethink their self-identity and change their life expectations and aspirations (Metcalfe et al., 2008).

One major component important in deciding when to disclose a child's at-risk status is whether or not a relative who is presenting symptoms is living nearby. If the child is exposed to a relative, such as a grandparent or a parent, who has developed HD it is more beneficial that the child be told about HD. Seeing first hand the destruction that HD can cause can be a very difficult thing to take. Without knowing what the disease is, how it works, and what it means to everyone in the family, the child can become more anxious and scared than if he or she were to know (Keenan et al., 2009).

An open communication style is most conducive to successful coping with the information about HD. This is defined as talking openly and honestly, allowing children to ask questions, helping them seek outside sources of support and information, and supporting their doing this on their own timeline (Coulson et al., 2007). Children are able to comprehend things a bit at a time, and as they understand more they have more questions. By keeping the lines of communication open the child will feel less stress and anxiety. The disclosure of at-risk status is also best if done gradually. Overloading children with information can leave them feeling confused and more anxious (Keenan et al., 2007).

Proposal/Recommendations

I recommend that parents who are at risk for HD tell their children about HD at an early stage in their children's lives. This is especially important for children who are exposed to a family member who are manifesting the symptoms of HD. These children should be told when they are as young as four years old to avoid unnecessary anxiety. Children who will not be exposed to HD first hand can be told a little later on, such as eight to ten years old, to still avoid some of the major adjustment issues. Parents rarely regret telling their children younger, while parents often regret telling their children later. Children need time to adjust to the information about the disease and ask questions. Locating face-to-face or online support groups can be helpful for additional support.

In addition, many parents felt as though they were not supported by medical professionals in the process of informing their children of their at-risk status. The process

of explaining a genetic disease to children has two additional difficulties outside of the psychosocial issues discussed above. First, many individuals who are at-risk have a hard time understanding what the disease is and how it works themselves which makes it difficult to explain to anyone else. Second, explaining a complex disease such as HD in a way that a child will understand is also difficult. To begin to address the problem of a lack of support, I have developed a pamphlet for parents to use when explaining HD to children. Parents can use it as an added resource, to help when disclosing information about HD. This pamphlet has been designed based on my recommendations that children be told early.

Limitations

Most of the research on which this recommendation is based on was completed in Scotland. Similar studies, on how young people learn and cope with HD, need to be repeated in the United States to show that the results can be generalized. Scotland and the United States are very different culturally, which could make the results different. Scotland is clan oriented, more so than the U.S. The Scottish do not move around as frequently as those in the U.S. Thus they are more likely to live close to a relative who shows symptoms. This may make the Scottish children more in need of learning of HD earlier.

Research is also needed to ascertain if using the resources provided aids parents in informing their children and aid children in coping and understanding HD. This research might also explore how medical professionals can be of use to parents and children in the

process. Currently parents do not feel as though they were helped by doctors or genetic counselors to prepare for talking with the children. Trying to establish a more active role for medical professionals in the disclosure process may be helpful for families during the difficult times. In addition, research could be done to look at additional variables (other than being close to an affected family member) that are considered when deciding whether to wait to inform.

Conclusion

Huntington's disease can cause many problems outside of its symptoms. It can cause individuals anxiety, stress, and pain even before any symptoms manifest. Those who are affected by HD are faced with many stressful and complex decisions, including whether to get tested, if they should reproduce, whom they should tell about HD, and what they should hope for in their lives. These decisions are harder to make when they are faced unexpectedly. If an individual always knew about HD, he or she would be able to start thinking about those decisions from an early age. The incorporation of HD into their identities is helpful in the coping process and can aid in decision making down the road. Telling children about their risk for HD may be a difficult process but it is better for them to learn early. Disclosure of at-risk HD status to children should be done early and openly. By allowing children to ask questions, learn about HD on their own timeline, and adjust to it over time they are able to cope more effectively.

Appendix

Pamphlet attached on the next page.

Explaining Huntington's Disease

Explaining the basics of Huntington's disease (HD) to those at-risk is one of the most important jobs of any family member. While it may be difficult to start, the knowledge will help those at risk make informed life decisions and cope more effectively later on. It is in the family's best interest to introduce HD to those who are at risk while they are still young. This process should be gradual, as parts of the disease are difficult to understand at first. Knowing that you are at risk does not make you grow up faster, but it helps you understand your family and your life more fully.

University of Northern Iowa
University Honors Program

This informational pamphlet was completed by Krysten Shipley as an addition to an Undergraduate Honors Thesis Project in conjunction with Dr. Ira Simet

Living At Risk Huntington's Disease

Explaining the basics of Huntington's Disease to those at risk

University of Northern Iowa
University Honors Program

Huntington's disease

To aid families in the process of explaining HD, this is a brief summary of Huntington's disease and the issues that are associated with the disease. It can be used as an aid in answering questions and beginning discussions.

AN OVERVIEW OF HD

In 1872, American physician George Huntington described a disease that can be traced back to the Middle Ages. This disease, now known as HD, is hereditary and progressive. HD's major symptoms are uncontrollable movements or shaking, behavioral changes, memory loss, and mood swings. The symptoms begin around the age of 40 and slowly worsen over time. Currently there are around 15,000 Americans living with HD and around 150,000 Americans living with a 50% risk.

HEREDITARY NATURE OF HD

Huntington's disease is passed from parent to child. This means that if an individual has HD, his or her children will have a 50% chance of also having HD.

PROGRESSIVE NATURE OF HD

Around the age of 40 the symptoms of HD will appear. Once the first symptom begins, the rest follow slowly. The disease builds and the symptoms get worse. In general, those who have HD will eventually need support in the form of a caregiver.

SYMPTOMS OF HD

The major symptoms of HD are chorea, mood swings, behavioral changes, and memory loss. Chorea is uncontrollable movements or shaking. This generally starts in the fingers, feet, face, or trunk. Individuals with HD also have trouble driving, learning new things, remembering a fact, or making decisions. As the disease progresses, concentration on intellectual tasks becomes increasingly difficult. Some individuals lose the ability to recognize family members, but most remain aware of their environment.

THE FUTURE

While currently there is no cure, researchers are very optimistic about future treatments. Knowing about HD and its impact on the family can help individuals cope effectively. Support from outside sources is available; contact your doctor to learn more about possible support groups.

ADDITIONAL RESOURCES

For more information and support visit:

National Institute of Health
www.ninds.nih.gov/disorder/huntington/detail_huntington.htm

Huntington's Disease Society of America
www.hdsa.org

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