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Huntington's Disease:
Ethical Dilemma of Family Relations and Personal Autonomy

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Abstract

Huntington's Disease (HD) is a late onset hereditary disorder that constitutes as a rare, progressive neurodegenerative disease. Due to its nature of being genetic, it brings about an ethical dilemma unique to its own situation. Based on life experience of a person whose family has been affected by the disorder for generations, this paper addresses various concerns about HD via the perspective of rehabilitation psychology. It tackles functional and psychosocial impacts of the disorder. In addition, personal autonomy and familial responsibility is discussed in relation to genetic testing. Conclusively, it outlines various multi-disciplinary treatments that may help increase quality of life for families afflicted with the disorder.

Keywords: rehabilitation, ethics, autonomy

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Huntington's Disease (HD) is a late onset hereditary disorder that constitutes a rare, progressive neurodegenerative disorder. It was previously known as Huntington's Chorea, and was named after physician George Huntington, who characterized the disorder. HD is an autosomal dominant genetic condition, meaning that a person only needs to inherit one copy of the mutated A gene from an affected parent to develop the disease. Each child of a parent with HD has a 50% chance of inheriting the mutation. In some cases, the mutation occurs spontaneously without a family history (Marcus, 2023).

The HTT gene present in chromosome 4 contains a triple repeat of nucleotides CAG which codes the amino acid glutamine. The triplet is usually repeated 10–35 times in a row, however people with Huntington's have this repeat more than 36 times which causes production of a faulty, misfolded huntingtin protein. This production of an abnormal huntingtin protein damages neurons in specific regions of the brain, particularly the basal ganglia and cerebral cortex. Over time, this neurodegeneration results in worsening motor dysfunction, cognitive decline, and psychiatric disturbances (Jiang et al., 2023).

Symptoms typically appear in adulthood between the ages of 30 and 50. The disease progresses slowly over many years and eventually leads to severe disability and the need for full-time care (Marcus, 2023). The length of the CAG repeat expansion also influences disease onset. Larger repeat expansions are typically associated with earlier symptom onset and more severe disease progression. In some families, the mutation becomes more severe across generations through a process known as genetic anticipation, that makes the symptoms appear earlier with each generation (McColgan & Tabrizi, 2018).

HD affects the basal ganglia which is responsible for motor control, action selection, habit formation, and reward-based learning and the cerebral cortex that coordinates conscious thought, memory, language, perception, and voluntary movement. This results in the disorder affecting all these domains. One of the hallmark features of HD is chorea, which refers to involuntary, irregular, dance-like movements affecting the face, torso, and limbs. As the disease progresses, individuals may also develop dystonia that is characterised by involuntary, sustained or repetitive muscle contractions alongside muscle rigidity, impaired balance, and abnormal eye movements. These motor

impairments often interfere with walking, coordination, and daily activities. Motor symptoms worsen over time and may eventually lead to difficulty swallowing (dysphagia), impaired posture, and frequent falls. In advanced stages, individuals may lose the ability to walk or perform basic self-care tasks independently (Marcus, 2023).

Van Duijn et al. (2018) highlight that psychiatric symptoms are extremely common in HD and may appear before motor symptoms become noticeable. These symptoms include depression, irritability, anxiety, apathy, and aggression. Due to these psychiatric symptoms and the progressive nature of the disease, individuals with HD are at a higher risk of suicidal ideation and suicide compared to the general population, with it being one of the leading causes of premature death in HD patients. Other common causes of death include complications like malnutrition, infections, and aspiration pneumonia, which develop as a direct result of HD symptoms (Solberg et al., 2018).

The prevalence of HD varies between different ethnicities with higher rates in individuals of Caucasian descent and lower in individuals of African or Asian descent. More recent epidemiological studies estimate global prevalence at approximately 4–5 cases per 100,000 people, highlighting differences based on genetic ancestry and regional diagnostic practices (Medina et al., 2022). As HD is a genetic condition, there is currently no method to prevent the mutation itself, however, early detection is possible through genetic testing, which can identify the presence of the HTT mutation before symptoms develop (McColgan & Tabrizi, 2018). Predictive genetic testing is often offered to individuals with a family history of HD, along with genetic counseling to help individuals understand the medical, psychological, and ethical implications of learning their genetic status (Van Lonkhuizen et al., 2023).

HD presents a unique intersection between neurology, genetics, psychology, and ethics. Unlike many medical conditions that emerge unexpectedly, HD can often be predicted decades before symptoms begin through genetic testing. This predictive ability creates complex ethical and psychological challenges for individuals and families who carry the possibility of the mutation.

Living With Genetic Risk

To understand how these concepts shape lived experiences, I interviewed a genetics professor whose family is directly affected. This conversation highlighted that living with genetic risk involves more than just medical uncertainty as it also shapes identity, career decisions, and ethical beliefs. Her second cousin is currently diagnosed with the condition, and several other relatives showed symptoms as well. One relative died after living with HD, and another family member eventually required full-time care due to severe disability. Despite these experiences, HD has become somewhat normalized within the family. The interviewee described how relatives have organized fundraising events, such as charity go-kart races, to support research and raise awareness.

Interestingly, the interviewee described her own emotional response to the disease as relatively calm and matter of fact. Rather than expressing ongoing fear or distress about the possibility that she may eventually develop HD, she spoke about it in a pragmatic way. Her response reflects an important concept known as adjustment to chronic illness. According to Dekker & De Groot (2018), adjustment models suggest that individuals adapt psychologically to long-term health threats by gradually incorporating them into their understanding of themselves and the world. Over time, what initially appears threatening may become a normalized background condition that individuals acknowledge without allowing it to dominate their identity

In the interviewee's case, HD influenced her career path. She explained that growing up around the disease is what inspired her decision to become a geneticist. She expressed that rather than distancing herself from the disorder, she pursued this career to better understand genetic mechanism and disorders. From a rehabilitation psychology perspective, this reflects the concept of meaning-making, in which individuals transform difficult experiences into sources of purpose and motivation (Park, 2010). The presence of HD in her family did not limit her aspirations but instead guided her toward a profession where she could better understand the biological processes behind genetic illness.

Identity development is another important concept relevant to individuals living at risk for HD. Rehabilitation psychology highlights how chronic illness can reshape

personal identity, particularly when individuals must integrate health-related uncertainty into their sense of self. Chronic illness frequently disrupts a person's sense of self, prompting a process of identity reconstruction in which individuals reinterpret their experiences and develop a meaningful identity that incorporates the illness (Clur & Barnard, 2025). For people who carry or may carry the HD mutation, identity may include elements such as being "at risk," having family members with the disease, or belonging to a community affected by genetic illness. In the case of the interviewee, her identity as a scientist and genetics professor appears intertwined with her family experience.

Ethics & Eugenics

At the same time, the interview highlighted the complex ethical challenges associated with predictive genetic testing. The interviewee has never been tested for the HD mutation, despite having a family history that places her at risk. The reason for this decision is rooted in an ethical dilemma involving family privacy. Her mother has chosen not to undergo genetic testing, and a positive test result for the interviewee would automatically reveal that her mother also carries the mutation. Out of respect for her mother's autonomy and right not to know, the interviewee has decided not to pursue testing herself.

This situation illustrates a central ethical issue in genetic medicine: genetic information is rarely individual. Unlike most medical tests, which reveal information about a single person, genetic testing simultaneously reveals information about biologically related family members. Stuttgen et al., (2020) recognize that these shared implications can create significant interpersonal stress within families. Individuals may feel caught between their own desire for knowledge and the need to respect the preferences of relatives who do not want to know their genetic status. It is an extremely nuanced situation that demands to be dealt with caution.

The interviewee emphasized that the right not to know is an important ethical principle in genetics. While she personally respects her mother's decision, she also acknowledged that other family members might choose differently. However, it is not necessary in their circumstance as she mentions that her younger brother does not really understand the logistics nor does he care to. The assumption is that whenever the

interviewee gets tested so will the brother as there is no urgency for risk assessment from his side.

This perspective reflects the principle of relational autonomy, which acknowledges that medical decisions are often influenced by family relationships, cultural expectations, and shared experiences rather than being purely individual choices. Another important theme emerging from the interview involves the concept of invisible risk and because HD often develops later in life, individuals who carry the mutation may appear completely healthy for decades. The interviewee noted that genetic risk can affect a person's self-concept even when the condition is not outwardly visible. People who know they may develop HD sometimes make life decisions based on this possibility, such as choosing to travel earlier in life, adjusting career goals, or deciding not to have children.

The interviewee herself stated that she has never wanted children, so the possibility of transmitting the mutation to offspring was not a major factor in her life planning. However, she acknowledged that many individuals with genetic risk for HD face difficult reproductive decisions. Some may choose not to have children, while others may pursue options such as preimplantation genetic testing to reduce the risk of transmission. These decisions intersect with broader ethical debates surrounding reproductive autonomy and concerns about eugenics (Emmerich & Gordijn, 2019). The interviewee made a point to distinguish between historical forms of eugenics, which involved forced sterilization and reproductive control and contemporary genetic decision-making, which is typically based on individual choice rather than coercion.

Quality of Life

The interviewee also highlighted the role of stigma and social misunderstanding. She explained that outsiders may not fully appreciate why individuals make certain life choices, such as declining parenthood or prioritizing travel and experiences earlier in life. These decisions can be influenced by an awareness that the disease may eventually limit mobility, independence, and lifespan.

The biopsychosocial model could help with understanding the intersection of the domains in this disorder. It is a foundational framework in rehabilitation psychology, which emphasizes that health conditions involve interactions between biological,

psychological, and social factors (Wade & Halligan, 2017). While the biological component of HD involves progressive neurodegeneration affecting movement, cognition, and psychiatric functioning, the psychological and social dimensions begin much earlier for individuals living at risk. The interviewee's experience demonstrates how uncertainty about genetic status can shape life decisions, identity development, and family relationships even in the absence of physical symptoms.

Overall, quality of life is a central concern for rehabilitation psychologists working with individuals affected by HD. Van Lonkhuizen et al. (2023) study examining quality of life in persons living with HD found that psychological and social factors often have as much influence on well-being as the physical symptoms of the disorder. Individuals with HD frequently report reduced quality of life due to depression, cognitive decline, emotional distress, and difficulties maintaining social roles such as employment and family responsibilities. The review suggests that quality of life in HD cannot be understood solely through the progression of neurological symptoms. Psychological factors play a major role in shaping how individuals perceive their well-being. Social circumstances, including caregiving demands, financial strain, and access to supportive resources, also contribute significantly to quality-of-life outcomes. These findings reinforce the importance of the biopsychosocial model.

Treatments

The interviewee emphasized that currently, there are no FDA-approved treatments that cure Huntington's disease, and researchers are primarily focused on slowing its progression. Even small improvements in cognitive functioning or physical ability could significantly enhance quality of life for people living with the condition. For many patients, extending the period during which they can live independently without requiring ventilators, wheelchairs, or full-time care, would represent a meaningful improvement in quality of life. As a result, many current treatment strategies focus on delaying the onset of symptoms or slowing neurodegeneration rather than eliminating the disease.

Evidence-based psychological treatments for HD include cognitive-behavioral therapy (CBT), supportive psychotherapy, and psychoeducation. Cognitive-behavioral therapy can help individuals manage depressive symptoms, anxiety, and stress associated with disease progression. CBT focuses on identifying negative thought patterns,

developing coping strategies, and improving emotional regulation (Silver, 2003). Although research specific to HD is limited, studies suggest that structured psychotherapy can improve mood and psychological adjustment in individuals with neurodegenerative disorders (McColgan & Tabrizi, 2018)

Another important intervention is family-based counseling and genetic counseling as an HD diagnosis affects multiple members of the same family. Genetic counseling helps individuals understand the implications of predictive genetic testing, family planning decisions, and emotional responses related to learning one's genetic risk (Paulsen et al., 2013). Counseling can also address anticipatory grief, uncertainty about disease progression, and family conflict related to caregiving responsibilities (Muñoz-Ortega et al., 2025).

Psychiatric medication may also be used to manage behavioral symptoms. Antidepressants such as selective serotonin reuptake inhibitors (SSRIs) are frequently prescribed to treat depression and anxiety. Antipsychotic medications may help manage irritability, aggression, and psychotic symptoms, while medications such as tetrabenazine or deutetrabenazine are commonly used to reduce involuntary chorea movements (McColgan & Tabrizi, 2018).

From a scientific perspective, developing treatments for HD is extremely challenging. The condition is caused by a toxic form of the huntingtin protein that progressively damages neurons in the brain. Although researchers understand that the mutant protein plays a central role in the disease, the normal biological function of the huntingtin protein is still not fully understood. Because the protein appears to be involved in essential cellular processes such as neuronal development and intracellular transport, completely removing or disabling the gene could produce harmful unintended effects (Zaheer et al., 2025). Therefore, many therapeutic strategies aim to reduce the production of the mutant huntingtin protein rather than eliminate the gene entirely.

Current research on HD increasingly focuses on gene-targeting therapies such as CRISPR, which aim to reduce or modify the expression of the mutant HTT gene responsible for the disorder. CRISPR technologies allow researchers to edit specific DNA sequences or alter RNA transcripts to decrease production of the toxic huntingtin protein

(Morelli et al., 2023). However, these strategies remain largely experimental and are not yet available for clinical use.

Motor-Cognitive Synergy

Rehabilitation plays a critical role in supporting individuals with HD by maintaining functional abilities and promoting independence as motor, cognitive, and speech functions gradually decline. Physical therapy is commonly employed to enhance balance, coordination, and mobility, with exercise programs including gait training, strengthening, and aerobic activity shown to reduce fall risk and support overall brain health and mood (Fritz et al., 2017). Occupational therapy focuses on preserving independence in daily activities, such as dressing, cooking, and personal care, often through adaptive equipment, task simplification, and environmental modifications that allow safe functioning at home and in the community. Speech-language therapy addresses dysarthria (a motor speech disorder caused by muscle weakness or paralysis) and dysphagia (difficulty swallowing). This intervention provides communication strategies, voice exercises, and swallowing protocols to minimize aspiration risk and improve verbal effectiveness (McColgan & Tabrizi, 2018). Finally, cognitive rehabilitation uses structured training to strengthen memory, attention, and executive skills. Although cognitive decline is progressive, these interventions help individuals maintain daily functioning and independence for longer periods (Fritz et al., 2023).

Rehabilitation psychologists should focus on promoting adaptive coping strategies, emotional regulation, and resilience. Interventions may involve helping individuals adjust to changes in independence, develop problem-solving skills, and maintain a sense of personal identity despite disease progression. Supporting family members and caregivers is also crucial, as caregiver stress and burnout are common in HD (Carney et al., 2025). In addition, rehabilitation psychologists should be aware of the ethical complexities surrounding genetic testing, stigma, and family planning decisions. Providing culturally sensitive care and advocating for equitable access to services can help ensure that individuals with HD receive appropriate psychological and rehabilitative support.

Overall, the interview highlights how genetic risk fundamentally shapes identity, career choices and family relationships. Addressing these interconnected biological, psychological, and social experiences is essential in navigating the ethical and emotional

challenges that emerge due to HD. Ultimately, while HD remains incurable, evidence-based psychological and rehabilitation interventions offer critical support designed to improve quality of life, preserve functional abilities, and help individuals and families as they adapt to the challenges of the disorder.

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