

Intricacies of Huntington's Disease: From Genetic Roots to Symptomatology

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DESCRIPTION

Huntington's disease, also known as Huntington's chorea, is a neurodegenerative disorder that affects the central nervous system. This disease gradually robs individuals of their ability to control movements, affects cognitive functions, and often leads to emotional disturbances. In this article, we will discover the intricacies of Huntington's disease, from its genetic roots to the impact it has on individuals and their families.

Huntington's disease is fundamentally a genetic disorder, stemming from a mutation in the Huntingtin (*HTT*) gene located on chromosome 4. The mutation involves an abnormal repetition of a specific DNA sequence called a CAG trinucleotide repeat. Typically, this sequence repeats between 10 and 35 times within the gene. However, in individuals with Huntington's disease, the CAG sequence is repeated 36 times or more. The expansion of the CAG repeat leads to the production of an abnormal form of the huntingtin protein, which plays a crucial role in the normal functioning of nerve cells. The mutated huntingtin protein has toxic effects on neurons, particularly those in the brain's striatum, leading to the characteristic symptoms of Huntington's disease.

Symptoms: The triad of motor, cognitive, and psychiatric manifestations

Huntington's disease is often characterized by a triad of symptoms, each manifesting in varying degrees and at different stages of the disease.

Motor symptoms: The term "chorea" in Huntington's chorea refers to the involuntary, dance-like movements that individuals with the disease often exhibit. These movements, known as chorea, can include jerking or writhing motions, making everyday activities challenging for those affected. As the disease progresses, coordination and balance are further compromised, leading to difficulties in walking and performing basic motor tasks.

Cognitive symptoms: Huntington's disease doesn't solely impact motor functions; it also inflicts cognitive impairments. Memory loss, difficulty concentrating, and a decline in problem-solving

skills are common cognitive symptoms. As the disease advances, individuals may experience significant cognitive decline, leading to challenges in daily life and a loss of independence.

Psychiatric symptoms: Psychiatric symptoms are a prominent aspect of Huntington's disease, contributing to the complex nature of the disorder. Depression, anxiety, and irritability are common, often exacerbated by the challenges posed by the physical and cognitive symptoms. Individuals may also exhibit impulsive behavior and personality changes, further complicating their emotional well-being.

Progression

Huntington's disease is progressive, with symptoms typically emerging in mid-adulthood, usually between the ages of 30 and 50. However, variations in the age of onset can occur, and in rare cases, symptoms may manifest in childhood or late adulthood. The progression of Huntington's disease can be divided into stages, each marked by specific symptoms and challenges. In the early stages, subtle motor symptoms may be mistaken for clumsiness, while cognitive and psychiatric symptoms may not be immediately apparent. As the disease advances, chorea becomes more pronounced, and cognitive decline becomes increasingly evident.

In the later stages of Huntington's disease, individuals often require assistance with daily activities, and motor functions are severely compromised. Swallowing difficulties and weight loss are common, posing additional health risks. The duration of the disease varies, with an average lifespan after the onset of symptoms ranging from 10 to 30 years.

Treatment and management

While there is currently no cure for Huntington's disease, various therapeutic approaches aim to manage symptoms, improve quality of life, and provide support for affected individuals and their families.

Medications: Certain medications, such as tetrabenazine, can help manage chorea and other motor symptoms. Antidepressants and antipsychotic medications may be

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prescribed to address psychiatric symptoms, providing relief from emotional distress.

Physical and occupational therapy: Physical and occupational therapy play crucial roles in maintaining mobility and independence for individuals with Huntington's disease. These therapies focus on improving muscle strength, coordination, and balance, enabling individuals to adapt to changing motor abilities.

Supportive care: A multidisciplinary approach involving healthcare professionals, including neurologists, psychologists, and social workers, is essential in providing comprehensive care. Support groups and counseling services can also offer emotional support to individuals and families grappling with the impact of Huntington's disease.

CONCLUSION

Huntington's disease, with its complex interplay of genetic factors and progressive symptoms, poses immense challenges for individuals and their families. Yet, amidst the difficulties, there is hope. Advances in research, coupled with the dedication of healthcare professionals and the resilience of affected individuals, offer glimpses of a future where the complexity of Huntington's disease may be unraveled, leading to improved treatments and, perhaps one day, a cure. Until then, a tapestry of confidence in the face of hardship is being created by the combined efforts of the scientific community and the constant encouragement of families and carers.