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Epigenetics Research: Open Access

Commentary

# Huntington's Disease: A Genetic Disorder

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## DESCRIPTION

Huntington's disease is a genetic disorder that affects the nervous system, causing a progressive decline in motor skills, cognitive abilities, and psychiatric functions. This disease is caused by a mutation in the Huntingtin (HTT) gene on chromosome 4, which leads to the production of a toxic protein called mutant Huntingtin (mHTT).

Huntington's disease is inherited in an autosomal dominant pattern, meaning that only one copy of the mutated gene is required to develop the disease. Individuals with a parent who has Huntington's disease have a 50% chance of inheriting the mutation and developing the disease themselves. The onset of Huntington's disease typically occurs between the ages of 30 and 50, but it can appear earlier or later in life.

Symptoms of Huntington's disease can vary widely between individuals, but they generally involve a combination of physical, cognitive, and psychiatric problems. The earliest symptoms may include mood swings, irritability, and depression, as well as mild motor problems such as clumsiness or unsteady gait. As the disease progresses, individuals may experience more severe motor symptoms, such as involuntary movements (chorea), difficulty with speech and swallowing, and problems with balance and coordination. Cognitive decline may also occur, leading to difficulty with memory, concentration, and decision-making.

While there is no cure for Huntington's disease, there are treatments available to manage its symptoms and improve quality of life. Medications can help to control chorea and other motor symptoms, while therapy and support groups can assist with emotional and psychological challenges. Genetic counseling can also help individuals and their families to understand their risk of inheriting the disease and make informed decisions about family planning.

One of the challenges of studying Huntington's disease is that the mutated HTT gene is very large and complex, with many different regions that can be affected by the mutation. Researchers are working to better understand the molecular mechanisms that underlie the disease, with the goal of identifying new targets for therapy. This work is complicated by the fact that the symptoms of Huntington's disease are very diverse, and different individuals may experience different combinations of symptoms depending on which parts of the brain are affected.

Research into Huntington's disease is ongoing, and recent advances in gene editing technologies such as CRISPR-Cas9 have raised the possibility of developing new treatments or even a cure. In 2020, the FDA approved the first drug specifically designed to slow the progression of Huntington's disease by targeting the mHTT protein. While this treatment is not a cure, it represents a significant step forward in the development of therapies for this devastating disease.

Despite these challenges, there is hope that continued research into Huntington's disease will lead to new treatments and ultimately a cure. In the meantime, individuals and families affected by Huntington's disease can find support and resources through organizations such as the Huntington's Disease Society of America and the Hereditary Disease Foundation. With increased awareness and understanding of this devastating disease, we can work together to improve the lives of those affected by Huntington's disease and their families.

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**Received:** 01-Mar-2022, Manuscript No. EROA-22-23557; **Editor assigned:** 03-Mar-2022, PreQC No. EROA-22-23557 (PQ); **Reviewed:** 17-Mar-2022, QC No. EROA-22-23557; **Revised:** 24-Mar-2022, Manuscript No. EROA-22-23557 (R); **Published:** 31-Mar-2022, DOI: 10.35248/EROA.22.4.109.

**Citation:** Drake M (2022) Huntington's Disease: A Genetic Disorder. J Epigenetics Res. 4:109.