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Editorial

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Editorial Note On Huntingtons Disease

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Huntington's Disease (HD), also known as Huntington's chorea, is a neurodegenerative disease that is mostly inherited. The earliest symptoms are often subtle problems with mood or mental abilities. A general lack of coordination and an unsteady gait often follow. As the disease advances, uncoordinated, involuntary body movements known as chorea become more apparent. Physical abilities gradually worsen until coordinated movement becomes difficult and the person is unable to talk. Mental abilities generally decline into dementia. The specific symptoms vary somewhat between people. Symptoms usually begin between 30 and 50 years of age but can start at any age. The disease may develop earlier in life in each successive generation. About eight percent of cases start before the age of 20 years, and are known as juvenile HD, which typically present with the slow movement symptoms of Parkinson's disease rather than those of chorea.

HD is typically inherited from an affected parent, who carries a mutation in the huntingtin gene (HTT). However, up to 10% of cases are due to a new mutation. The huntingtin gene provides the genetic information for huntingtin protein (htt). Expansion of CAG repeats of cytosine-adenine-guanine (known as a trinucleotide repeat expansion) in the gene coding for the huntingtin protein results in an abnormal mutant protein (mhtt), which gradually damages brain cells through a number of possible mechanisms. Diagnosis is by genetic testing, which can be carried out at any time, regardless of whether or not symptoms are present. This fact raises several ethical debates: the age at which an individual is considered mature enough to choose testing; whether parents have the right to have their children tested; and managing confidentiality and disclosure of test results.

There is no cure for HD, and full-time care is required in the later stages. Treatments can relieve some symptoms and, in some, improve quality of life. The best evidence for treatment of the movement problems is with tetrabenazine. HD affects about 4 to 15 in 100,000 people of European descent. It is rare among Japanese, while the occurrence rate in Africa is unknown. The disease affects men and women equally. Complications such as pneumonia, heart disease, and physical injury from falls reduce life expectancy. Suicide is the cause of death in about 9% of cases. Death typically occurs 15–20 years from when the disease was first detected. Symptoms of Huntington's disease most commonly become noticeable between the ages of 30 and 50 years, but they can begin at any age. Their progression is often described in early stages, middle stages, and late stages with an earlier prodromal phase. In the early stages, there are subtle personality changes, problems in cognition, and physical skills, irritabilty, and mood swings, that may all go unnoticed. The physical symptoms are usually the first to be noticed. Almost everyone with HD eventually exhibits similar physical symptoms, but the onset, progression and extent of cognitive and behavioral symptoms vary significantly between individuals.

The first likely description of the disease was in 1841 by American physician Charles Oscar Waters. The condition was described in further detail in 1872 by American physician George Huntington. The genetic basis was discovered in 1993 by an international collaborative effort led by the Hereditary Disease Foundation. Research and support organizations began forming in the late 1960s to increase public awareness, provide support for individuals and their families and promote research. Research directions include determining the exact mechanism of the disease, improving animal models to aid with research, testing of medications to treat symptoms or slow the progression of the disease, and studying procedures such as stem-cell therapy with the goal of replacing damaged or lostneurons.

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