

HUNTINGTON'S DISEASE (HD)

Huntington's disease (HD) is a fatal disease typically characterized by involuntary movements (chorea) and cognitive decline (dementia). It is caused by a genetic mutation that can be passed down from generation to generation. HD is an illness with profound neurological and psychiatric features.

Huntington's disease affects certain structures deep within the brain, particularly the basal ganglia, which are responsible for important functions, such as movement and coordination. Structures responsible for thought, perception, and memory are also affected, likely due to connections from the basal ganglia to the frontal lobes. As a result, patients may experience uncontrolled movements (such as twisting and turning), loss of intellectual abilities, and emotional and behavioral disturbances.

Demographics

In the United States, the overall prevalence of HD is about 1 in every 10,000-20,000 persons. The disease begins most often in mid-adulthood, although the age of onset varies greatly. Typical age of onset is between 30 and 55 years old, however juvenile onset occurs in about 10% of families.

Symptoms

The early signs of the disease vary greatly from person to person. In general, the earlier the symptoms appear, the faster the disease progresses.

Early symptoms include personality changes, such as mood swings, irritability, apathy, depression, anger, and even aggression. Early in the disease, cognitive decline manifests as memory and learning difficulties, judgment impairment, trouble with driving, answering questions, or making decisions. As the disease progresses, concentration on intellectual tasks becomes increasingly difficult.

Manifestations of chorea appear at various points of the disease, depending on the patient, and begin as uncontrolled movements of the extremities, face, or trunk that become progressively worse. These may be preceded by fidgety movements, restlessness, clumsiness, or imbalance; the person may appear uncoordinated and may even fall.

The disease can progress to the point where speech is slurred and vital functions, such as swallowing, eating, speaking, and especially walking, continue to decline. Some patients are unable to recognize others. Many, however, remain aware of their environment and are able to express emotions.

The duration of the disease from the time the first symptoms appear ranges from 10 to 30 years. Most common causes of death are infection, fall-related injuries, and other complications.

Pattern of Inheritance

HD has an autosomal-dominant pattern of inheritance, which means that only one copy of the mutated gene, from either parent, causes the disease. A parent with the gene that causes HD has a 50% chance at each pregnancy of passing the gene to their offspring. Males and females are equally affected. The gene for HD (located on chromosome 4), when activated, makes the protein huntingtin, which accumulates in abnormal amounts in the brains of HD patients. The mutation in HD consists of repeats of a trinucleotide codon (CAG). How exactly the defective protein causes harm in HD is not completely understood.

Individuals who inherit the HD gene may develop the disease earlier and in a more severe form than the previous generation. This is termed “anticipation.”

Very rarely, an individual can develop HD who has no known family history of the disorder. Cases like this are thought to occur due to spontaneous mutations or from a missed diagnosis in the previous generation.

Diagnosis

In 1993, the identification and location of the HD gene was discovered. This has made it possible to determine who will develop the disease through genetic testing. This is done by obtaining a blood sample and analyzing the DNA. An accurate family history continues to be important. Brain imaging studies, such as computed tomography (CT) and magnetic resonance imaging (MRI), may show atrophy of the affected parts of the brain, especially the caudate nuclei and putamen (parts of basal ganglia). Additionally, generalized brain atrophy can also be seen. However, genetic testing is the only way to make a definite diagnosis of HD while the patient is living, either to predict eventual onset or confirm the cause of symptoms.

Treatment

Currently, there is no cure for HD or ability to slow it down. However, there are treatments available to help control the symptoms. Antipsychotic drugs may help to alleviate involuntary movements and may also be used to help control hallucinations, delusions, and violent outbursts. Antipsychotic drugs, however, can have severe side effects, including stiffness and sedation, and for that reason are used in the lowest possible doses.

Antidepressants are used for depression and tranquilizers can help with severe mood swings. Studies are underway to determine if antioxidants and other agents may provide neuroprotection, and therefore prevent degeneration in HD. Thus far, these studies have not been shown to be beneficial.

Caregivers

Due to the debilitating nature of the disease, caring for a patient with HD can be physically and emotionally exhausting. As the disease progresses and patients become unable to perform activities of daily living (ADLs), they require increased amounts of care. Weight loss, not due to decreased caloric intake, is a common feature of patients with HD. In later stages of the disease, patients are often bed-bound and have significant swallowing problems. Special devices to assist in ADLs, special diet to aid in swallowing, and increasing calories to counteract weight loss may eventually require consideration.

If you live in the San Francisco Bay Area, we offer an **HD Support Group** for family members or caregivers.