Huntington’s Disease

Huntington’s disease (HD) is an inherited neurodegenerative disease that generally results in uncontrolled movements, emotional disturbances, and mental deterioration. HD is characterized by a combination of motor, cognitive, and psychiatric symptoms that evolve over many years. The survival time after the onset of symptoms can range from 10 to 30 years. Currently, there is no known cure for HD.

Prevalance of Huntington’s Disease

- In the United States, about 25,000 people have HD. Each child of a parent with the disease has a 50 percent chance of inheriting the HD gene and will ultimately develop the disease.
- HD does not discriminate based on ethnicity or sex: while the disease occurs throughout the world, there are geographic clusters that do exist.

Signs and Symptoms of Huntington’s Disease

- The signs and symptoms of HD most often appear in middle age, though the age of onset varies from person to person.
  - In general, the earlier symptoms appear, the faster the disease will progress.
  - Early signs of the disease vary from person to person, but initial symptoms of HD often include uncharacteristic mood swings, irritability, apathy, passivity, depression, or anger, which can increase or decrease as the disease progresses.
  - For some individuals, chorea associated with HD may be their most prominent symptom. Chorea is present in an estimated 90 percent of people with HD at some point in their illness.
- HD may affect the individual’s judgment, memory, or other cognitive functions.
  - Early signs of cognition problems include having trouble driving, learning new things, remembering a fact, or making a decision. Concentration typically worsens as the disease progresses.
- As chorea associated with HD progresses, the involuntary, jerk-like movements worsen and cause patients to develop a characteristic dance-like motion.
- In advanced stages of HD, patients may develop severe dementia and progressive motor dysfunction, which may lead to difficulty walking. They also may suffer from poor dietary intake, difficulty speaking, and they eventually may not be able to take care of themselves.
  - Complications may result from injuries related to serious falls, poor nutrition, infection, choking, or heart failure.

Genetics of Huntington’s Disease

- HD results from genetically programmed degeneration of brain cells called neurons.
- HD is a hereditary disease that is passed from parent to child through a mutation in a normal gene.
  - Each parent has two copies of every chromosome, but gives only one copy to their child. As a result, each child of an HD parent has a 50 percent chance of inheriting the HD gene.
  - If the child of an HD parent does not inherit the HD gene, he or she will not develop the disease and cannot pass it to subsequent generations.
- A person who inherits the HD gene and lives long enough will eventually develop the disease.
Diagnosing Huntington’s Disease

- HD is typically diagnosed by a neurologist after a thorough neurological examination, including brain imaging or genetic testing, and a review of family history.³

- The discovery of the HD gene in 1993 resulted in a genetic test to confirm an HD diagnosis in an individual who is exhibiting HD-like symptoms.⁹

- Presymptomatic testing is also available for people who have HD in their family, but are not yet exhibiting symptoms.³
  - Genetic testing for the HD mutation can be particularly useful when there is an unknown or negative family history.⁸

For more information on Huntington’s disease, please visit:

- The Hereditary Disease Foundation, www.hdfoundation.org
- Huntington’s Disease Society of America, www.HDSA.org

SOURCES

1 Mayo Clinic.com Web site: Huntington’s Disease mayoclinic.com/print/huntingtons-disease/DS00401/DSECTION=all&METHOD=print Last accessed 9/22/2011


