

Huntington disease (HD) is an inherited disease that causes certain nerve cells in the brain to die. People are born with the gene that causes HD, but symptoms don't usually appear until mid-adulthood. As HD progresses, physical, emotional and cognitive functioning will be affected as the person with HD becomes less able to control movements, recall events, make decisions and control emotions.

About Huntington disease

HD is a monogenetic hereditary neurodegenerative disease caused by a defective gene on chromosome 4. If a parent has HD, their children will have a 50% chance of inheriting the gene. The HD gene is responsible for producing a protein called huntingtin, a protein that is found throughout the body's tissue but that is most concentrated in the brain. While the normal function of the huntingtin protein is still unknown, the defective form has been found to cause HD.

HD causes nerve cells, called neurons, in parts of the brain to gradually deteriorate. The parts of the brain that are most affected by HD are the outer layer of the brain, known as the cortex, and the deeper areas of the brain, called the basal ganglia which includes the caudate and the putamin.

Since the cortex is responsible for higher level cognitive functioning like thinking and understanding, and the basal ganglia helps people to perform smooth movements, HD generally affects both the physical and cognitive functioning.

Other names historically used for HD include Chorea, Chronic Progressive Chorea, Huntington's Chorea and Hereditary Chorea. The word "Chorea" (a Greek term for dance) is used to describe the involuntary movements that people with HD commonly experience.

What are the symptoms?

The age of symptom onset and the rate of the disease progression vary among people living with HD. However, symptoms of HD typically begin in people between the ages of 30 and 50, and usually progress over a 10 to 25 year period.

Early symptoms of HD often include subtle cognitive changes. Slight physical changes may also develop at this stage - a person may experience an increased difficulty controlling their movements.

As the disease progresses, it is not uncommon for a person with HD to encounter personality changes such as irritability, depression and mood swings. A person with HD may experience trouble with memory, concentration, learning new things or making decisions. Obsessive-compulsive behaviour, such as continually repeating the same activity, is also a common feature of HD. The initial physical symptoms will gradually develop into more obvious involuntary movements such as jerking and twitching of the head, neck, arms and legs.

The later stage symptoms of HD may include an increased difficulty with concentrating, walking, eating independently and swallowing.

How is Huntington disease diagnosed?

A thorough assessment will often include physical, neurological and psychiatric exams and a review of the person's complete family medical history to help rule out other conditions. Changes in behaviour and personality, like increased irritability, can be mistaken for other conditions, causing some of the early symptoms of HD to be initially overlooked.

People may undergo genetic testing via a blood test to confirm or rule out HD if they are exhibiting symptoms of the disease. Brain imaging (MRI) may be requested by a physician to detect any structural changes in the parts of the brain that are affected by HD.

What is predictive testing?

Predictive genetic testing is also available for adults who are not currently showing signs of HD, but who have a strong family history of the disease. In this case a result of gene positive or gene negative is given, but a diagnosis of HD is not given until symptoms develop – usually much later in life. Prior to going through predictive testing, it is important that the person speak with a genetic counselor to discuss both the advantages and disadvantages of genetic testing.

What are the causes or the risk factors?

Huntington disease is a familial disease, passed from parent to child through a mutation in the normal gene that is responsible for the huntingtin protein. Anyone with a parent with HD has a 50 percent chance of inheriting the gene, and everyone who inherits the gene will eventually develop the disorder. In about 1 to 3 percent of cases, no history of the disease can be found in other family members.

Is there treatment?

Medications can help manage symptoms, but cannot slow down or stop the disease and there is currently no cure for HD. On a promising note, there are several drug trials underway, which may bring new treatment approaches forward.

Therapeutic approaches are also supporting people living with HD to manage symptoms of their disease. Occupational therapy can help improve the functional ability of people living with HD through the use of assistive devices, while physical therapy can help maintain physical abilities. Since HD can impair the muscle control needed for eating and speaking, speech therapy can help improve a person's verbal communication and can address eating and swallowing challenges.

It is important that caregivers and professionals work together to help manage the most effective treatment for each individual, since the disease develops differently in different people, even within the same family or generation.

Support is available:

Visit the Alzheimer Society's website at www.alzheimer.ca or contact your local Alzheimer Society. For more information on HD, please visit Huntington Society of Canada at www.huntingtonsociety.ca.

Additional resources:

- HD Buzz – for HD information and research updates: <http://en.hdbuzz.net/>
- Alzheimer’s Society UK: http://www.alzheimers.org.uk/site/scripts/documents_info.php?documentID=135
- Mayo Clinic:
<http://www.mayoclinic.org/diseases-conditions/huntingtons-disease/basics/complications/con-20030685?p=1>
- National Institute of Neurological Disorders and Stroke:
<http://www.ninds.nih.gov/disorders/huntington/huntington.htm>

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