Huntington's Disease
Factsheet

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What is Huntington's disease?

Huntington's disease (HD) is an hereditary disorder of the central nervous system. It used to be known as Huntington's Chorea or HC. HD causes a very wide range of symptoms in both men and women and usually develops in adulthood (people aged 30 -50) but can affect children and teenagers (Juvenile HD).

George Huntington

(April 9, 1850 – March 3, 1916) An American physician who contributed a classic clinical description of the disease that bears his name, he wrote a medical paper based around his observations of the hereditary disease in multiple generations of a family in East Hampton on Long Island.

Chromosome No 4

Humans have 46 chromosomes (23 pairs). The faulty gene that causes Huntington's disease is found on chromosome number four.

The normal copy of the gene produces a protein called huntingtin, but the faulty gene is larger than normal and produces a larger form of huntingtin.

Scientists don't yet understand the normal function of huntingtin protein or how a few dozen extra repeats in its genetic blueprint lead to the devastating symptoms of Huntington's disease.

Huntingtin

Cells in parts of the brain – specifically, the basal ganglia and parts of the cortex – are very sensitive to the effects of the abnormal huntingtin. This makes them function poorly and eventually die. The brain normally sends messages through the basal ganglia. When this part of the brain is damaged, it causes problems with control of movement, behaviour and thinking. It is still unclear exactly how abnormal huntingtin affects the brain cells and why some are more sensitive than others.

Pre-clinical HD

This phase is called the preclinical or prodromal phase and relates to those who have been tested as HD positive but are some years away from being officially diagnosed as having HD.

They are likely to show some attributes of suffering from HD such as:

- Lower emotional recognition
- Increased mood swings
- More likely to suffer depression or be more irritable
- Higher risk of suicide due to depression

Affects 12 people in every 100,000

Main categories

People with HD may express a wide variety of symptoms, which are typically grouped into three main categories.

- Movement
- Cognitive
- Psychiatric

Stages of HD

- Pre-clinical HD
- Early stage HD
- Middle stage HD
- Late stage HD
- End of life

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Early stage HD

This is the earliest time that symptoms become noticeable enough to warrant a diagnosis.

Some symptoms – particularly cognitive and behavioural symptoms – may make it harder for people to work and perform at their usual level. However, at this stage, people are still able to maintain a fairly normal lifestyle and can generally continue to work, drive, and live independently.

Motor symptoms usually begin in the extremities of the body; people experience involuntary twitches in their fingers, toes, and face. Onlookers generally don't notice these motions, or assume that they're just nervous twitches. People in the early years of HD also experience a subtle loss of coordination, and may have more trouble performing complicated motions.

Middle stage HD

By the middle stage of HD, people often lose their ability to work and drive, and might be unable to perform household chores. Eating can become challenging, as patients have trouble performing the complicated series of muscle movements needed to swallow. Speech becomes slurred, and walking becomes staggered. However, many people are still able to eat, dress, and take care of hygiene with some help. Physical therapists can help patients control their voluntary movements; speech pathologists can help patients deal with swallowing and speaking; occupational therapists can help patients deal with changes in their thoughts.

Late stage HD

By the late stage of the disease, people with HD require help in all aspects of life. They are generally unable to speak, and remain bedridden. Since it becomes more and more difficult to care for a patient as the disease progresses, patients often spend the last few years of life in a nursing home. Choking is a major concern; it becomes extremely difficult to swallow, so most late-stage patients need to be fed with a tube that is inserted surgically into the stomach or small intestine. Many patients have trouble urinating or become constipated, and some patients have trouble sleeping normally.

End of life

Ultimately, people with HD die an average of 10-20 years after symptoms begin. Death is believed to be primarily from complications of the disease. The most common complication is aspiration pneumonia, in which patients get a fatal infection from unintentionally inhaling a piece of food. Suicide is the second leading cause of death.

There is a 50:50 chance of inheriting the faulty gene.

Effects

- Uncontrollable muscular movements
- Difficulty swallowing
- Weight loss
- Stumbling or clumsiness
- Difficulty concentrating
- Memory loss
- Psychological problems
- Mood swings
- Aggressive behaviour

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