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# What is Huntington Disease?

**A brief description**

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## What is HD?

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Huntington disease (HD) is an inherited brain disorder. It was named after the doctor who first described it in 1872, George Huntington, and it used to be called Huntington's chorea.

HD causes cells in specific parts of the brain to die: the caudate, the putamen, and, as the disease progresses, the cerebral cortex. The caudate and putamen have connections to many other areas of the brain, and help to control body movement, emotions, thinking, behaviour, and perception of the world.

As the brain cells die, people with Huntington's become less able to control movements, recall recent events, make decisions, and control emotions. The disease leads to incapacitation and, eventually, death.

## Who gets it?

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Approximately one in every 10,000 Canadians has HD, and approximately five in every 10,000 are at risk of developing the disease.

Huntington disease is a genetic disorder, caused by a faulty gene on chromosome 4. The HD gene is dominant, which means that each child of a parent with HD has a 50% chance of inheriting the disease and is said to be "at risk". Males and females have the same risk of inheriting the disease.

Huntington's occurs in all races, but it is most prevalent in people of European ancestry.

Primarily, HD affects adults. Symptoms usually appear between the ages of 30 and 45, but the disease can first appear in children as young as five, or in adults in their 70s.

## What causes it?

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It's not clear how the abnormal HD gene causes the disease. Since the discovery of the gene in 1993, scientists have been working hard to discover the biochemical processes that causes the brain cells to die. So far, we have learned that the HD gene produces a protein called "huntingtin". In people with HD, this protein gets cut into one short piece and one longer piece. The shorter pieces stick together to form a "protein ball". Scientists are currently investigating whether it is the breakage of the protein, the formation of protein balls, or some other process that leads to cell death. They are also trying to understand why only certain brain cells die.

## What are the symptoms?

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There are three main types of symptoms in Huntington disease:

- *physical symptoms*, including involuntary movements and clumsiness
- *emotional symptoms*, including depression, irritability and obsessiveness
- *cognitive symptoms*, including loss of ability to recall information, loss of attention and difficulty with decision making.

There is a lot of variation in symptoms, and not every person will have all the symptoms to the same degree. For example, some people with HD may have a lot of difficulty with involuntary movements, while others may have fewer physical symptoms but many emotional or cognitive difficulties. Symptoms also vary with each stage of the disease.

## Early stage

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Early symptoms appear as slight physical changes, and may also include cognitive or emotional changes.

A clinical diagnosis of HD is based on the presence of involuntary movements. These may initially consist of “nervous” activity, fidgeting, a twitching in the hands or feet, or excessive restlessness. Individuals may notice a little clumsiness, changes in handwriting, or difficulty with daily tasks such as driving.

In addition to the physical symptoms of HD, there are often subtle cognitive signs as well. People with early Huntington’s may find they have difficulty organizing routine matters or coping effectively with new situations. Difficulty recalling information may make them appear forgetful. Work activities may become more time-consuming, and decision-making and attention to details may be impaired.

Early emotional symptoms may be equally subtle. Individuals at this stage may experience more periods of depression, apathy, irritability, or impulsiveness, or perhaps changes in personality.

At this stage, people with Huntington’s can function quite well at work and home.

## Intermediate stages

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As the disease progresses, the symptoms become worse. The initial motor symptoms will gradually develop into more obvious involuntary movements such as jerking and twitching of the head, neck, arms and legs. These movements may interfere with walking, speaking and swallowing.

People at this stage of Huntington’s often look

as if they're drunk: they stagger when they walk and their speech is slurred. They have increasing difficulty working or managing a household, but can still deal with most activities of daily living.

## **Advanced stages**

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The advanced stages of Huntington's typically involve fewer involuntary movements and more rigidity. People in these stages of HD can no longer manage the activities of daily living, and they usually require professional nursing. Difficulties with swallowing, communication and weight loss are common.

Death usually occurs 15 to 20 years after the onset of the disease. People don't die from Huntington's itself, but from complications such as choking, heart failure, or infection.

*(Adapted from Dr. Ira Shoulson's five-stage model of Huntington disease.)*

## **Juvenile HD**

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About ten percent of Huntington's cases are considered "juvenile"—the symptoms appear in childhood or adolescence.

The symptoms of juvenile HD are somewhat different from the adult disease. Children with HD move slowly and stiffly, they have increasing difficulty learning, and they can have convulsions or epileptic seizures. Some children have severe behavioural problems.

The earlier the disease onset, the more likely the child is to be very rigid and appear very different from the majority of adult cases. This often makes it difficult to diagnose. Children with HD are more likely to have an affected father than an affected mother.

## How is it diagnosed?

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Huntington's is usually diagnosed using neurological and psychological tests. Sometimes doctors use brain scans to see whether the caudate and putamen are working properly, or they use the genetic test (see below) to confirm the diagnosis.

## Genetic testing

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Since 1993, genetic testing for HD has been available. This means people who are at risk for Huntington's or who believe they have the symptoms can take a blood test to tell them whether they have the gene that causes HD.

Genes are made up of DNA, and DNA molecules consist of four bases known as A (adenine), T (thymine), G (guanine) and C (cytosine). The gene that causes HD has a region in which a sequence of three bases (CAG) is repeated many times.

New information about the HD gene continues to emerge, but it is currently thought that the normal version of the gene has 35 or fewer CAG repeats. A gene with 36 or more repeats is associated with Huntington disease, although there is an intermediate range which may or may not lead to the onset of symptoms. About one percent of people tested fall into this intermediate category.

Many people at risk choose not to take the test. One reason may be that there is still no treatment to prevent HD from developing if the gene is present. But some people want to know whether they will eventually get the disease, so they can make plans about careers, insurance, family planning, and other

issues. And others just want to know.

Anyone considering taking the test should have genetic counselling. This will ensure that he or she understands what the possible outcomes could be, and whether the decision to be tested is the right one at that time.

The fact there is a test available that will predict whether someone will develop HD raises a number of important issues. These include insurance, employment, family planning, and they can affect whether someone decides to undergo genetic testing.

## **Are there any treatments?**

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At the moment there are no treatments that will slow down or stop the disease. However, there are some drugs available that can reduce some of the symptoms, such as depression, anxiety, and involuntary movements. These drugs can have side effects, so not everyone with Huntington's uses them.

Neurologists, psychologists, genetic counsellors and social workers can play an important role in helping individuals or families deal with the disease. Physical therapists, occupational therapists and speech therapists can also help people with Huntington's cope better with some of the symptoms. And because people with HD often lose a lot of weight, a nutritionist can be very helpful.

There is a great deal of research going on to find a cure for HD, and scientists are optimistic that there will soon be a breakthrough. By investigating the biochemical mechanisms of cell death in HD, they hope to be able to develop new drugs that will slow, stop, or even reverse the course of the disease.

Other researchers are looking at surgical treatments, such as implanting fetal brain cells into the brains of Huntington's patients in the hope the cells will grow and take over the functions of the dead cells.

There are also various drug trials underway under the auspices of the Huntington Study Group, an international consortium focussed on clinical research in Huntington disease.

## **Huntington Society of Canada**

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The Huntington Society of Canada is a national network of volunteers and professionals working to find new treatments and ultimately a cure for Huntington disease, and to improve quality of life for people with HD and their families.

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