

# What is Huntington Disease?

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Huntington disease (HD) is an inherited brain disorder. HD causes cells in parts of the brain to die: specifically the caudate, the putamen and, as the disease progresses, the cerebral cortex. As the brain cells die, a person with Huntington's becomes less able to control movements, recall events, make decisions and control emotions. The disease leads to incapacitation and, eventually, death (generally due to other health complications).

## Who gets it?

Huntington disease is a genetic disorder. 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. Symptoms usually appear between the ages of 30 and 50, but the disease can appear in children or seniors.

## What causes it?

It is 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 cause 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.

## Are There Treatments?

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

On a more promising note, there are several advanced drug trials underway under the auspices of the Huntington Study Group, an international consortium focused on clinical research in Huntington disease.

Researchers are also 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.

Researchers feel we are close to reliable treatments; the urgency lies in educating as many people as possible, including new outreach efforts in rural and culturally diverse communities.

## Clinical Trials

HSC continues to partner with leading organizations that are conducting and preparing for clinical trials. Individuals have played an essential role in all aspects of Huntington disease (HD) research. It was family history records that guided the discovery of the HD gene mutation and blood samples from families around the world that made genetic research possible. HSC bridges the relationship between researchers and individuals by educating Canadians on the importance of the clinical trial process; how they can get involved; and why their participation is so crucial. It will be through clinical trials that new research breakthroughs will be moved from discovery to the HD community.

To move clinical trial research forward more people are needed. For a clinical trial to be successful a wide participation rate is required in order to find meaningful results. Canadians are quickly becoming the highest per capita participants in HD clinical trials in the world, but we are not there yet. HSC plays a key role in bridging the relationship between researchers and individuals by educating Canadians on the importance of the clinical trial process; how they can get involved; and why their participation is so crucial. It will be through clinical trials that new research breakthroughs will be moved from discovery to the HD community.



## How is HD Diagnosed?

Huntington's is usually diagnosed using neurological and psychological tests, and with a review of family history. Sometimes doctors use brain scans to see whether the specific parts of the brain, primarily the caudate and putamen are working properly, or genetic testing is completed to confirm the diagnosis.

## The Huntington Society of Canada

Family is at the heart of our community. Our families and volunteers tell a powerful story of caring people who pull together to improve quality of life for Canadians impacted by HD. At the Huntington Society of Canada (HSC), we understand what you are going through, whether you have the disease yourself, are caring for someone who does, are gene positive, or are at-risk of inheriting HD. We are a not-for-profit charitable organization that raises funds to deliver counselling services and other supports to individuals and families living with Huntington disease (HD). The Society works with health and social services professionals to enable them to better serve people living with HD. We also fund medical research leading to treatments that will delay or stop the progression of the disease.