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 the quality of life for of Canadians impacted by HD. At the Huntington Society of Canada (HSC), we understand what you're 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 which 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.

Huntington Disease FACTS

- HD is a fatal hereditary brain disorder
- A child born to a parent with HD has a 50% chance of sharing the same fate
- 1 in every 1,000 Canadians is directly or indirectly impacted by HD
- Currently there is no cure for HD

Huntington Society of Canada
HUNTINGTON
Société Huntington du Canada

FOR MORE INFORMATION, OR TO DONATE VISIT:
huntingtonsociety.ca OR CALL **1-800-998-7398**

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What is HUNTINGTON DISEASE?

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WHAT is Huntington disease?

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 45, but the disease can appear in children or seniors.

WHAT are the symptoms?

- Emotional turmoil (depression, apathy, irritability, anxiety, obsessive behaviour)
- Cognitive loss (inability to focus, plan, recall or make decisions; impaired insight)
- Physical deterioration (weight loss, involuntary movements, diminished coordination, difficulty walking, talking, swallowing)

There are significant variations in symptoms, and not every person will have all the symptoms to the same degree. Symptoms also vary with each stage of the disease.

Early Stages

Early symptoms of the disease often include subtle cognitive changes.

- May have difficulty organizing routine matters or coping effectively with new situations
- May have difficulty recalling information may make them appear forgetful
- Work activities may become more time-consuming
- Decision making and attention to details may be impaired
- May include irritability

Slight physical changes may also develop at this stage. There can be involuntary movements which may initially consist of "nervous" activity, fidgeting, a twitching of the hands or feet, or excessive restlessness. Individuals may also notice a little awkwardness, changes in handwriting, or difficulty with daily tasks such as driving. At this stage, people with Huntington's can function quite well at work and at home.

Intermediate Stages

As the disease progresses, the symptoms become worse. The initial physical symptoms will gradually develop into more obvious involuntary movements such as jerking and twitching of the head, neck and arms and legs. These movements may interfere with walking, speaking and swallowing. People at this stage of Huntington's often stagger when they walk and their speech may become slurred. They may have increasing difficulty working or managing a household, but can still deal with most activities of daily living.

Advanced Stages

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 usually require professional nursing. Difficulties with swallowing, communication, and weight loss are common.

Death usually occurs 15 to 25 years after the onset of the disease. People don't die from Huntington's itself, but from complications such as choking, heart failure, infection or aspiration pneumonia.

Juvenile HD

Close to 10 percent of Huntington's cases are considered "juvenile" – that is, the symptoms occur in childhood or adolescence. Symptoms of juvenile HD are somewhat different from the adult disease.

- Children with HD move slowly and stiffly
- Increased difficulty learning
- May have convulsions or epileptic seizures
- Some children have severe behavioural problems

Because these symptoms can be very different from those in adults, it can be difficult to diagnose. 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. It is important that all of these professionals work together to help manage the most effective treatment for each individual, since the disease often develops differently in different people.

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?

In 2011, Canadian researchers provided bold new hope when they were able to successfully reverse the physical symptoms disease in a mouse model. This groundbreaking research is the first of its kind in the world and represents a major milestone on the path to the discovery of an effective treatment. Testing and further study of this new model of prevention are ongoing.

At the moment, however, 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.

Scientists are extremely excited about the hope that these drugs may hold. 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.

Genetic Testing

Since 1986, genetic testing for HD has been available; however, a direct test for the disease was developed in 1993. This means people who are at risk for Huntington's or who believe they have the symptoms can take a blood test to determine whether they have the gene that causes HD. Many people at risk choose not to take the test. It is a personal decision and varies from person to person as there is still no treatment to prevent HD from developing if the gene is present. Others make the decision to be tested so they can make arrangements as far as careers, family planning, and other issues are concerned. Anyone considering taking the test should have genetic counselling. This will ensure that the person understands what the possible outcomes could be, and whether the decision to be tested is the right one for them, at that time.

Genetic DISCRIMINATION

It is critical to create a safe environment for people with hereditary diseases, like Huntington's, and ensure they are free to come forward, get the help they need and participate in clinical trials. This is why HSC advocates on behalf of genetic fairness. Canada is the only G8 country that does not have protection over the use of its citizens' genetic information. As the leading organization of the Canadian Coalition for Genetic Fairness (CCGF), HSC is dedicated to establishing protection over the use of personal DNA information for all Canadians.