WHAT IS HD?

Huntington disease (HD) is a hereditary, neurodegenerative illness with physical, cognitive and emotional symptoms. Huntington disease is caused by a mutation in the gene that makes the protein called huntingtin. In people with HD, the CAG sequence is repeated too many times at the beginning of the gene. That causes cells to manufacture a harmful protein called mutant huntingtin. The mutant huntingtin protein causes certain parts of the brain to die - specifically the caudate, the putamen and, as the disease progresses, the cerebral cortex. As the brain cells die, symptoms will appear in each of the three components (physical, cognitive and emotional).

To date, there are no drugs to slow or stop the progression of Huntington disease; however, there are specific drugs available to reduce some of the symptoms. Research is being conducted in Canada and globally to find promising treatments and approaches to treating HD.

Who Gets HD?

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. HD occurs in all races. Symptoms usually appear between the ages of 35 and 55, but the disease can appear in youth (under 20 years – Juvenile HD) or older adults (Late Onset HD).

Symptoms of HD

Symptoms vary from person to person and at different stages of the disease.

- Physical symptoms: weight loss, involuntary movements (chorea), diminished coordination, difficulty walking, talking and swallowing
- Cognitive symptoms: difficulty with focus, planning, recall of information and making decisions; impaired insight
- Emotional symptoms: depression, apathy, irritability, anxiety, obsessive behaviour

Early Stages of HD

At this stage, people with HD can function quite well at work and at home. Early symptoms of the disease may include the following:

- Difficulty organizing routine matters or coping effectively with new situations
- Decreased ability to recall information and make decisions
- Increased difficulty with work activities
- Decreased attention to details
- Mood changes and irritability
- Minor involuntary movements (e.g. "nervous" activity, fidgeting, a twitching of the limbs or excessive restlessness)
- Changes in handwriting or difficulty with daily tasks such as driving

Intermediate Stages of HD

People with HD in the intermediate stage may have increasing difficulty working or managing a household but can still deal with most activities of daily living. Symptoms progress over time and may include the following:

- More obvious involuntary movements (chorea)
- Increased difficulty with walking, coordination and balance
- Challenges with speaking (speech may become slurred) and delays in thinking process
- Solving problems becomes more difficult
- Difficulties with swallowing
- Weight loss

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Advanced Stages of HD

People in the advanced stages of HD can no longer manage the activities of daily living and usually require professional care. Symptoms will include the following:

- Decrease in involuntary movements and increase in rigidity
- Increased difficulties with swallowing
- The ability to communicate diminishes, but understanding what is being said remains possible
- Significant weight loss

Juvenile HD

About 10% of people diagnosed with Huntington disease have the juvenile form. In Juvenile Huntington Disease (JHD), the symptoms occur in childhood or adolescence (before the age of 20) and tend to follow a more rapid course. Diagnosis of JHD is very difficult because the symptoms of Juvenile HD have somewhat different features from the adult form of the disease. Chorea (involuntary movements) is a much less prominent feature and may be absent altogether. Initial symptoms may include the following:

- Slow and stiff movement (rigidity), and sometimes tremors
- Difficulty learning in school and attention deficits
- Increase in responsive behaviours
- Seizures

Having a team of professionals to work with the person with JHD and the family is very important.

Late Onset HD (Diagnosis after age 60)

There is a wide range in the age of disease onset for people with HD. If a diagnosis is received after age 60, it is considered Late Onset HD. Knowledge of the typical age of onset (ages 35 to 55) sometimes leads physicians to miss the diagnosis, because doctors incorrectly believe the person is too old to develop HD. Late onset consists of about 10% of all HD diagnoses.

RESOURCES

Ongoing support, education and information is available from the Huntington Society of Canada (HSC). You can find a listing of our Family Services team members at www.huntingtonsociety.ca/family-services-team.

A Physician's Guide to the Management of Huntington Disease

Understanding Behaviour in Huntington Disease: A Guide for Professionals

A Carer's Guide for Huntington Disease

HSC educational modules (especially Responsive Behaviours, Caregiver Overview, Thinking Module)

Fact sheets on a variety of other topics including Cognitive Changes in HD, Have you Met HD?, Responsive Behaviours, Tips When Working with Individuals with HD are available at www.hdfactsheets.ca.

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