

Celebrating 40 years!



Huntington Society of
Canada's 40th Anniversary
Campaign:

Grassroots to Mountaintops.
Building a new chapter in our
history starts now.

The Grassroots to
Mountaintops campaign goal
is to raise \$5 million to meet
critical needs.

With the help of our strategic
partners and supporters, the
Huntington Society of Canada
continues to be an
innovator by creating
opportunities across Canada.

To learn more click here.

Enroll!

Updates from the Enroll-HD
global community



Enroll-HD is a very exciting
global initiative that is a
planning and consultation
process for clinical trial
registration for Huntington
disease.

Publications and fact sheets
are available on the HSC
website in the following
categories: Individuals
with HD; Those At-Risk;
Caregivers; Families;
Professionals.

www.huntingtonsociety.ca

Strength & Knowledge

July 25, 2013
Volume 6, Issue 4

Facts on Genetic Counselling

What is Huntington Disease?

Huntington disease is a genetic neurodegenerative disorder, affecting approximately 1 in 7,000 Canadians. The condition is progressive in nature and over time, as brain cells are affected, individuals are less able to control movements, make decisions and control emotions.

What causes Huntington Disease?

Genes are paired units of hereditary information found inside each cell in the body that direct our growth, development and function. We inherit one member of each gene pair from each parent. 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 Huntington disease is called the HD gene. There is a region in the HD gene in which a sequence of three bases (CAG) is repeated many times. For individuals with Huntington disease, the CAG sequence has increased (expanded) into a range considered abnormal or disease-causing. An HD gene expansion is passed on in families in a dominant manner (50% chance for both male and female children to inherit the expansion).

What is Genetic Testing for Huntington Disease?

Genetic testing involves the examination of an individual's DNA which is obtained from a blood sample. Testing is done in a specialized laboratory to determine the number of CAG repeats in both copies of the HD gene. As everyone has two copies of the

HD gene, it is common for a person to have two different CAG repeat sizes (one inherited from their mother and one inherited from their father). Genetic testing can take several weeks to be completed. Test results for Huntington disease can be complex and it is recommended that they are reviewed with a genetics specialist in order to fully understand what it means for an individual and their family.

Genetic testing can serve two purposes:

1. **Diagnostic Testing:** done to confirm a diagnosis of Huntington disease in individuals who have symptoms of the disease.
2. **Predictive testing:** refers to a process whereby an individual, with a family history of Huntington disease, wishes to know whether or not they have inherited the expanded HD gene, even though they currently have no symptoms.

What is Genetic Counselling?

The goal of genetic counselling for Huntington disease is to help individual better understand the impact of the testing results for them, their partner and their family. Anyone considering genetic testing should have genetic counselling. There is no fee for genetic counselling or genetic testing (if deemed appropriate) as it is covered by provincial health insurance. In some provinces, for individuals who live further from a medical genetics clinic, some or all sessions may be able to be done by videoconference to a local hospital.

Continued on page 2

EHDN's Standards of Care for HD patients

*"The most comprehensive effort so far to document
best practice in the clinical management of HD".*



EUROPEAN HUNTINGTON'S DISEASE NETWORK

What is the Predictive Testing Process for Huntington Disease?

Predictive testing is available through medical genetics clinics in most major centres in Canada. This process may vary somewhat between centres, but usually consists of multiple appointments with genetic and psychosocial counselling and a blood test. In some centres there may be a neurological exam. The time involved to complete the predictive testing protocol can vary from several weeks to several months.

What Should I Consider?

The decision to undergo genetic testing is a very personal one and many people with a family history of Huntington disease choose not to take the test. On the other hand, some people want to know whether they will eventually develop symptoms of the condition so that they can make plans regarding careers, insurance, family planning and other issues.

The following might be some questions that would be addressed with a genetic counsellor:

- What are the possible results of my genetic test?
- What would the result mean for me, my partner and my family?
- If I choose not to proceed with predictive testing right now, can I start the process again at a later date?
- Who should I share my genetic test result with? Parents? Spouse? Children? Employer? Anyone?
- How do I share my genetic test result?
- Are there issues of 'genetic' discrimination?
- Can a positive genetic test result impact my ability to get insurance?
- What are my reproductive and family building options?
- What do I need to know about research in HD?
- If my test shows that I will develop symptoms, what resources are in my local area (i.e. neurology, Huntington Society of Canada contact, etc.)?
- Where do I go if I need additional help and support?

What are the Possible Genetic Test Results?

Normal or "Gene Negative": The number of CAG repeats is 26 or less. This individual will not go on to develop Huntington disease and their children are not at increased risk either.

Intermediate: The number of CAG repeats is between 27 and 35. This individual will not go on to develop Huntington disease but in some cases may pass on an expansion to their children because the CAG repeat can be unstable when being passed from one generation to the next. This can mean that sometimes children will be at higher risk for developing the condition.

Reduced Penetrance: The number of CAG repeats is between 36 and 39. Individual with a CAG repeat size in this range may not develop any symptoms of Huntington disease, even if they live until old age. However, this result also means that the next generation may be at risk of inheriting a larger expansion as it would be unstable as described above.

Full Penetrance or "Gene-Positive": The number of CAG repeats is 40 or more. This individual will go on to develop symptoms of Huntington disease at some point in their life. It does not necessarily mean that they have signs of the condition already but it predicts that they will have symptoms at some point. The result does not provide information as to the exact age an individual will be when they start to have symptoms or what those symptoms will be.

Unaffected		Affected	
Normal <26 CAG	Intermediate 27-35 CAG	Red. Penetrance 36-39 CAG	Full Penetrance >40 CAG

How Do I Arrange for Genetic Counselling?

Anyone with a personal or family history of Huntington disease and who is of the age of majority is eligible for genetic testing. Most genetics clinics require a referral from your family doctor (or another healthcare professional involved in your care). To find a genetics clinic near you, contact Huntington Society: www.huntingtonsociety.ca or you can visit the Canadian Association of Genetic Counsellors website: <https://cagc-accg.ca/>.

More Information

For more information and personal perspectives on predictive genetic testing for Huntington disease see: <http://predictivetestingforhd.com/> and visit the Huntington Society of Canada's website (listed above) for other related topics.



HUNTINGTON'S DISEASE
YOUTH ORGANIZATION

HSC is a proud supporter of HDYO an International organization to help young people who are affected by HD. Visit www.HDYO.org

Staying Connected

Please visit our **website** and fill out our confidential contact information form to receive email communications from HSC. According to your preferences, we'll communicate with you in confidence by email about research findings, clinical trials, best practices of care, local and national events and happenings, volunteer opportunities, etc.



Follow us on Facebook and Twitter for daily updates!

Huntington Society of Canada
151 Frederick Street, Suite 400
Kitchener, ON N2H 2M2
1-800-998-7398
www.huntingtonsociety.ca