# GENETIC TESTING AND HUNTINGTON DISEASE (HD)

Living with a family history of Huntington disease (HD) can raise difficult questions about the future. Some people want to know whether they will develop HD so they can plan, while others choose not to test at all. Both choices are valid and deeply personal. The information in this factsheet is meant to help people at risk and their families understand genetic testing options and consider what might be right for them.

## Deciding Whether to Have Genetic Testing

- Genetic testing is a personal choice, and there is no right or wrong decision.
- Some people choose not to be tested
- Others want information to help plan for:
  - Career or education
  - Finances and insurance
  - Family planning
  - Long-term care and support
- Testing can be done now or later, or not at all
- Taking time to think and talk with professionals is encouraged

## Possible Genetic Test Results

Genetic testing looks at the number of CAG repeats in the HD gene, which helps explain risk.

## HUNTINGTON DISEASE CAG REPEATS

| Repeat Size (CAG) number | Size category | Affected with HD? | Risk for children? |
|---|---|---|---|
| Up to 26 | Normal | Will NOT develop HD | Children NOT at-risk |
| 27 – 35 | Intermediate | Will NOT develop HD | Small chance for children to have HD if repeat size gets bigger |
| 36 – 39 | Reduced Penetrance | Some will develop HD (often later). Some will not | 50% chance of inheriting a repeat size with disease 'potential' |
| 40 or more | Affected (Full mutation) | WILL develop HD within normal lifespan | 50% chance of inheriting disease |


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- Repeat sizes outside the normal range (i.e., >26 CAG) can be unstable. It is possible they may get bigger (expand) when passed on to the next generation.
- Repeats are more likely to get bigger (expand) when passed from an affected father (paternal transmission). It does not mean this WILL happen, there is just a higher chance the repeat size will change.
- Larger repeat sizes are associated with earlier age of onset. Individuals with juvenile HD (onset <20yrs) usually have 60 or more CAG repeats.
- Repeat size is important but is likely not the only factor influencing age of onset. People with the same repeat size can have onset of symptoms several years apart.

## Why People Choose Genetic Testing

- Genetic testing can be used for different reasons, depending on a person's situation
- Diagnostic testing confirms HD in someone who already has symptoms
- Predictive testing is for people without symptoms who want to know if they inherited the gene

## What Is Genetic Counselling?

Genetic counselling helps people understand genetic testing and its possible impact before and after testing. Genetic counselling:

- Is required before predictive genetic testing
- Helps individuals prepare emotionally and practically
- Is covered by provincial health insurance
- May be offered in person or by video in some provinces
- Anyone considering genetic testing should meet with a genetic counsellor

## Topics Discussed with a Genetic Counsellor

Genetic counselling provides a safe and supportive space to ask questions, learn about HD, and think through decisions. Meeting with a genetic counsellor does not mean a person is expected or required to proceed with genetic testing. Many people seek genetic counselling to understand Huntington disease better, including how it is inherited and what living at risk may mean for themselves or their family. For some individuals, discussing the possible benefits and challenges of genetic testing is important, but testing does not need to be the main reason for connecting with genetics services.

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**Topics may include:**

- The process of genetic testing
- Possible test results and what they mean
- Family history review
- Exploring reasons for or against testing
- Impact on partners, children, and family members
- Whether testing can be delayed or stopped
- When and how results are shared
- Protecting privacy and preventing discrimination
- Insurance and financial considerations
- Family planning options – for Prenatal testing, see the Family Planning Fact Sheet
- Inheritance and genetics (CAG repeats) of HD
- Experience of HD within the family
- Research opportunities

## How to Arrange Genetic Counselling

- Accessing genetic counselling is the first step in the testing process. Genetic counselling:
- Is available to adults with a personal or family history of HD
- Usually requires a referral from:
  - Family doctor
  - Neurologist
  - Another healthcare provider
- Local HSC Family Services staff can help with referrals

Factsheet developed by the HSC Family Services team. Reviewed by Tanya Greaser, BA, BSW, Registered Social Worker and Family Services Educator, and David Macgregor, MSc, CCGC, Genetic counsellor, Provincial Medical Genetics Program, St. John's, Newfoundland

## SUPPORT AND RESOURCES

**Support is available whether or not you choose genetic testing.**

Canadian Association of Genetic Counsellors | www.cagc-accg.ca

Reach out to your local Resource Centre Director (RCD) of the Family Services Program at HSC for ongoing support and education at hdsupport.ca

If you are ready to receive individual or group support from the Huntington Society of Canada, you can self-refer here: http://contactme.cloud/form/huntingtonsociety

Informational fact sheets for family and friends impacted by HD are available to view, print and download at hdfactsheets.ca

Healthcare professionals and caregivers needing a more in depth understanding on caring for someone with HD can find guides at hdresources.ca



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