

# Predictive Testing & Diagnosis

## How do I know if I or someone I know is at risk of HD?

If you are a child of a parent with Huntington's disease (HD), then you have a 50% chance of inheriting the disease. The following are suggested steps to take if you suspect you or someone you know is at risk of HD.

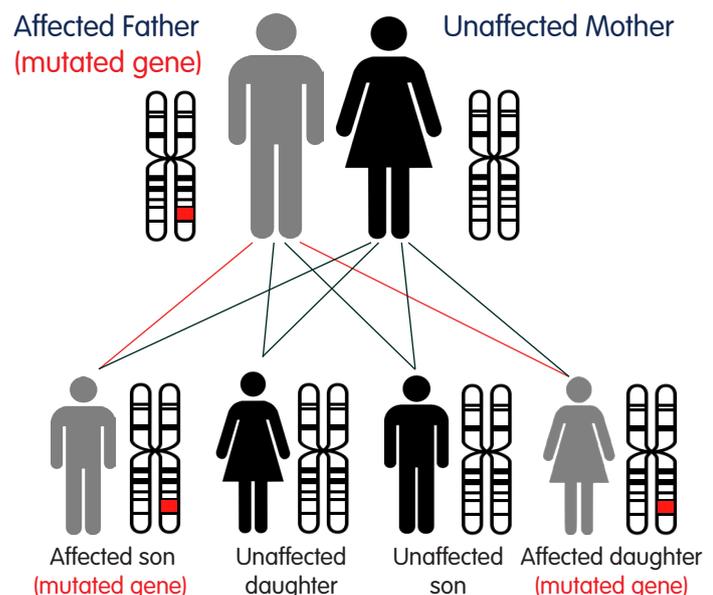
1. **Explore your family tree**, talk to your parents and/or relatives and ask questions regarding your family history.
2. **Contact your local HD association**. These are confidential support services that can provide you with information and suggest questions to ask medical professionals in relation to HD.
3. **Speak to your GP** and encourage them to contact your HD association for more information on how you can be assisted.

## The Genetics of HD

Our body consist of 23 pairs of chromosomes, 22 pairs of autosomes and 1 pair of sex chromosomes. Chromosomes are made of long strands of genes which are the basic unit of inheritance. We inherit one chromosome from our mother and one from our father (see diagram). The HD gene is located on chromosome 4.

People with HD have one 'working' copy of the gene and one 'faulty' copy. When they have children, they may pass on the 'working' copy or the 'faulty' one, giving each child a 50% chance of inheriting the faulty gene.

If you inherit the faulty gene from your affected parent, at some stage, you will develop the symptoms of HD. If you inherit the working gene, then you won't get the disease and none of your children will inherit the gene and develop HD.



## What is predictive testing?

Predictive testing is performed by extracting your DNA from a blood sample to identify whether you are carrying the faulty Huntingtin gene.

Predictive testing is generally available if you're 18 and over through local genetic services (please check with your local association on the location of these centres). The testing usually consists of genetic counselling, a neurological examination and a blood test. The time between the initial blood test and receiving your results is generally 6 to 8 weeks. Please refer to our website for referral processes.

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## It's a Personal Choice

There is no right or wrong decision whether to undergo predictive testing, it is a personal choice. Before making a decision:

- **Consider all relevant information** so you can make an informed choice (i.e. around future career and family planning, insurance, etc.)
- **Consider seeking advice** from a professional to help understand all possible outcomes and make sure you're emotionally ready to make the decision.
- If you decide to go through with the test, speak to your local HD association about **follow up support**, regardless of test result.

Please note that if you wish to have support through this process, Huntington's Victoria can attend these appointments with you.

## Diagnosis

In order to receive a diagnosis, you will need to see a HD specialist and obtain a referral from your GP. Consultation(s) with a specialist is covered by Medicare. The initial consultation may include:

- Discussion of family history
- Your current knowledge of HD
- Discussion of symptoms you believe to be exhibiting
- Neurological/cognitive examination (body movement, reflexes, memory etc.)
- Assessment of mental and emotional state



After the above assessments, the specialist will inform you of whether or not they believe you are exhibiting symptoms of HD. The specialist will require a blood test to confirm that your symptoms are associated with HD (if you have not already been through predictive testing).

## For further information:

Science of predictive testing & personal experiences <http://en.hdyo.org/eve/articles/53>

Genetic Health Services Victoria <http://www.vcgs.org.au/clinical/Default.aspx?docid=301fc9c2-4c51-4752-897a-9a8b0166aaa4>

Monash Health Genetics <http://www.monashhealth.org/page/Genetics>

Royal Melbourne Neurogenetics <https://www.thermh.org.au/health-professionals/clinical-services/genetic-medicine>