Saint Mary's Hospital
Manchester Centre for Genomic Medicine

Information for Patients

Predictive testing for the Huntington's Disease gene

Who is this leaflet for?

This leaflet is for individuals with a family history of Huntington's disease (HD). The leaflet has been designed to explain the steps involved in having a predictive gene test for HD.

What is a predictive gene test?

A predictive test provides information about whether or not a person who has a family history of HD will go on to develop the disease in the future. As the individual does not have symptoms themself, this type of test is called a predictive test (or a pre-symptomatic test). If someone already has symptoms, this would be called a diagnostic test.

What are the symptoms of HD?

HD is a slowly progressive illness, which means that the early symptoms are usually very subtle and it may be many years before the illness becomes a serious problem. HD is usually diagnosed when an individual has involuntary movements. However, other symptoms, such as poor memory and mood changes, may occur before the involuntary movements. Symptoms usually start to develop between the ages of 30-50, however in some cases it may be earlier or later than this.

What happens at my first appointment?

You can request a referral to the Genetics service through your GP. Your first meeting provides an opportunity for you to meet a member of our team and for us both to get more information. We will draw out a family tree to help work out who is at risk in the family. It is also important that we confirm the diagnosis in the family, as many inherited neurological conditions share similar features, and we need to ensure that we are testing for the right condition.

We can also discuss the clinical symptoms of HD and how it is inherited. For individuals who are well themselves and not showing signs of a condition, we would not offer the test at the first appointment. The first appointment will give you chance to explore the possible effect a result may have on you and your family.

How many appointments would I need?

Testing usually involves at least three appointments, but it is important to take as long as you need before deciding to go further with testing. You are able to withdraw from the testing process at any time. We understand that the process can seem very long, but it is based on the experiences of people who have been through predictive testing in the past, and the things they have found most helpful.

When would I get the test result?

If you decide to go ahead with predictive testing, a small blood sample would be taken. The test takes around 4 weeks, and we would arrange to see you in clinic to give you the result. We never give results over the phone, or in a letter, and we ask that you bring someone else with you when you come for your result. The staff involved will not know the result of your test beforehand, so you can telephone us at any time, without worrying that we know something and are not telling you.

Would I ever be declined the test?

It is your decision whether or not to have the test. We are here to give you information, to help you think through the issues before having the test and to help you prepare for the result if you decide to be tested. According to current guidelines, the test is only routinely available to those over the age of 18.

Are there any financial implications to the test?

Currently, a predictive test should not affect your ability to take out new insurance policies. An agreement (moratorium) is currently in place until 2021, which means that insurance companies should not ask for results of genetic testing (except for very large policies). More information is available from the HDA website (details at the end of this leaflet).

Is the test result 'clear cut'?

Everyone has a HD gene that contains 'CAG' repeats. It is the number of CAG repeats that determines whether someone will develop HD or not. Under 27 CAG repeats is always normal, whereas 39 repeats or more is abnormal and would cause HD at some stage. However, there is also the possibility of an 'intermediate' result. A repeat length of 27-35 is normal, but there is a small risk that the repeat may increase in future generations. A HD gene with 36-39 repeats is abnormal, but there is a chance that the person may be affected very late in life or even not at all.

What happens after the test result?

We would normally contact you by telephone a few weeks after the test result, and will offer you an appointment. Both 'gene positive' and 'gene negative' results can raise unexpected emotions and issues. If you carry the HD gene, you will also be offered annual appointments, and we will keep you up to date with research projects that may be relevant to you.

For more information

If you need more advice about any aspect of HD, you are welcome to contact:

Manchester Centre for Genomic Medicine
Sixth Floor
Saint Mary's Hospital
Oxford Road
Manchester
M13 9WL

Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145

The department is staffed Monday–Friday, 9.00 am to 5.00 pm.

Website: www.mangen.co.uk

The Huntington's Disease Association (HDA) produces a range of information leaflets that you might find helpful, and can be obtained by contacting the association at:

Huntington's Disease Association (HDA)
Suite 24
Liverpool Science Park
Innovation Centre 1
131 Mount Pleasant
Liverpool
L3 5TF

Telephone: 0151 331 5444
Fax: 0151 331 5441
Website: www.hda.org.uk
E-mail: info@hda.org.uk

The following websites also provide information, support and resources about HD:

www.predictivetestingforhd.com (written specifically for people considering a predictive test)
www.hdbuzz.net (provides information about HD research, written in plain language)
www.en.hdyo.org (aimed at younger people)

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).