



**Huntington's Disease Association**

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# **Huntington's disease**

## **Predictive Testing**

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**Fact Sheet**

## Predictive Testing for Huntington's Disease

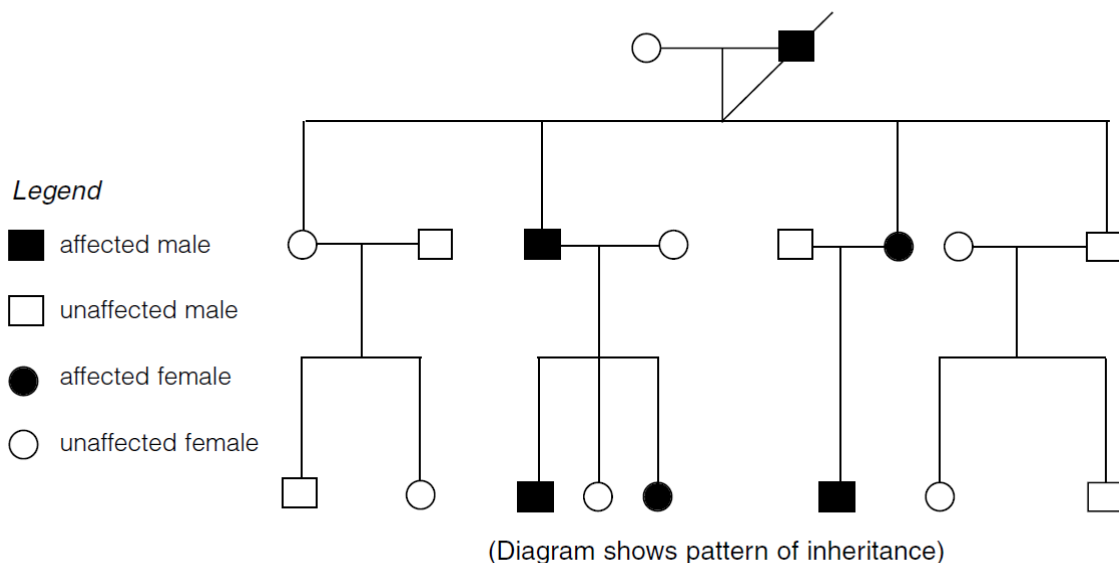
Huntington's disease is an hereditary disorder of the central nervous system. It is caused by a faulty gene. If one of your parents or another relative has Huntington's disease then you will probably be wondering about the likelihood of developing the disease yourself.

This fact sheet explains:

- How Huntington's disease is passed on
- The likelihood of developing the disease
- Genetic testing
- The Huntington gene
- Having children – including foetal exclusion testing

### How Huntington's disease is passed on

In each cell of your body you have 22 pairs of chromosomes (autosomes) and 2 sex chromosomes. One of each pair of autosomes comes from your father and one from your mother. Chromosomes contain genes which are the basic units of inheritance. The gene for Huntington's disease is on chromosome 4.



The gene for HD is larger than normal. Your affected parent has one normal sized copy of the gene and one larger sized copy. You got half your genetic material from each parent so it's a 50:50 (or 50%) chance that you got the half with the normal sized gene and a 50:50 (50%) chance that you got the half with the large size. There is a 50:50 chance each time, so each child is at 50% risk. Sometimes this is called a '1 in 2' chance of developing the disease.

If you have the faulty gene you will, at some stage, develop the disease. If you inherit the 'good' gene, you won't develop the disease and cannot pass it on to your children.

The symptoms of Huntington's disease usually develop when people are between 30 and 50 years old (though they can start much earlier or much later). The faulty gene is present from conception.

As the age of onset is so variable, an individual who carries the faulty gene may die before the disease has had time to develop and the relatives, then, may not know the true extent of their risk.

## **The likelihood of developing the disease**

If one of your parents has Huntington's disease your 'risk factor' does not remain at 50% all your life. As you get older, the likelihood of developing the disease decreases.

If you reach 70 years of age without any symptoms your risk will be very low. However, these are standardised figures and your Genetic Clinic will advise you individually of your risk.

The following table illustrates how the risk decreases over time for someone who is not symptomatic and who has a parent affected by HD:

Age	Percentage risk		Age	Percentage risk
20	49.6%		50	31.5%
25	49		55	24.8
30	47.6		60	18.7
35	45.5		65	12.8
40	42.5		70	6.2
45	37.8		75	4.6

For technical reasons, the figures used by some genetic clinics or in other countries may vary slightly from those shown in this leaflet.

It does not make any difference to your risk factor if:

- you look like your affected parent
- your brothers or sisters develop the disease
- you are male or female

## **Genetic Testing**

Living with the knowledge that you are at risk can be very worrying. You may feel that you would prefer to know for certain whether or not you have the faulty copy of the gene. A DNA test can now be carried out which will usually give you this information. In a few cases the test is still uninformative. Although the test is available, it does not mean that you should have it. You need to consider very carefully whether the test is right for you. If you do decide that it is, you need to think about things like its affect on life insurance, job opportunities etc. It is probable that a positive test result would make it more difficult to get life insurance.

If you have only just discovered that you are at risk, then be careful not to rush into making a decision. Once you have been given your test results, you can't change your mind about whether or not you wanted to know.

Only you can make the decision about whether you want to be tested and you usually need to be over eighteen years of age before it will be performed.

Parents, partners and other family members may pressure you one way or the other, but it remains your decision. Please let the Huntington's Disease Association know about any pressure which you feel is being put on you by health care professionals, employers or insurance companies.

If one of your grandparents has/had Huntington's disease but your own parent is so far not showing any symptoms and does not wish to be tested, you will need to talk things over particularly carefully. If you take the test and find that you have the faulty gene, then it will automatically mean that your parent has as well. Trying to keep this result secret is likely to be difficult or even impossible. So you will need to discuss how any adverse effects can be minimised. Though this is a very difficult issue the general opinion at the moment is that your right to have the test is greater than the right of your parent not to know.

Testing is only available at Regional Genetics Clinics, which are located throughout the country. A list of these centres is given at the end of this leaflet. You can ask your GP to arrange an appointment for you.

However not all GPs are aware that testing is now available so you may have to explain the situation. It might be useful to give your GP a copy of this fact sheet and other information from the HDA.

Going to the Genetics Clinic does not mean that you are obliged to take the test but it does give you the chance to talk over all the implications and any other concerns you may have. Each Clinic follows an agreed counselling procedure or 'protocol' which is usually spread over at least three sessions, to help you decide. You can withdraw from the procedure at any time. According to national and international guidelines, follow-up counselling – after you have been given the test result – should be available.

If you decide to have the test done you may have two separate blood samples taken (to double check the results). Your affected parent's blood may also be tested to check the original diagnosis of Huntington's disease. The DNA which is extracted from the blood is then analysed in a specialised laboratory. Some people find waiting for the results very stressful. If you feel there is a particularly long delay, do ask the Genetics Clinic for the reason. Technically, the test can be quite complicated both to perform and interpret. The Clinic will want to allow time to ensure that a result is ready for you and this may be from 4-6 weeks after the third counselling session.

## **The Huntington's Gene**

Genes are made up of DNA (deoxyribonucleic acid). DNA itself is made up of four chemicals which are known by letters of the alphabet:

A Adenine    G Guanine

C Cytosine    T Thymine

One section of the Huntington's gene contains three of these chemicals – CAG – repeated a number of times. In the faulty gene these three chemicals are repeated many times, like a 'molecular stutter'.

Four types of results are recognised:

Under 27 repeats is unequivocally normal. Between 27-35 repeats is normal, but there is a small chance that the repeat may increase in future generations. Between 36-39 repeats the result is abnormal but there is a chance the person may be affected very late in life or even not at all. Over 40 repeats is unequivocally abnormal.

Though the test can tell whether you carry the Huntington's disease mutation, it cannot tell you when the disease itself will start to develop.

## **Having Children – Including Prenatal Testing**

Knowing that you are at risk may affect your decisions about having a family of your own.

Some people decide never to have children at all, whilst others go ahead on the grounds that the children are likely to have many years of normal life before developing the disease (if they get it at all).

Couples at risk to Huntington's disease may find it more difficult to adopt although they may be able to undertake fostering. IVF (in vitro fertilisation) and AID (artificial insemination by donor) may also be considered.

Your decision to have children may depend upon the results of genetic testing. If testing shows that you don't have the faulty gene, then you can't pass it on to your children.

If you do have the faulty gene then your unborn children can be tested to see if they have inherited it. If you do not know if you have the faulty gene, and do not want to take the test yourself there is a different type of prenatal test which can be performed using linkage analysis. It was used before the gene was found and direct testing was available so it is not as accurate but it does not alter your risk.

If you are considering this option do discuss it at the Genetics Clinic well before embarking on the pregnancy.

Pre-implantation Genetic Diagnosis (PGD) offers another alternative to testing for HD in a pregnancy (prenatal testing). PGD gives a couple the chance of conceiving a pregnancy that should be unaffected by HD. PGD involves the couple undergoing IVF treatment (fertility treatment) even if they are a normally fertile couple. These embryos are then tested for HD before they are implanted in the woman's womb. Only embryos without the HD mutation are chosen for replacement. The hope is then that the couple will be successfully pregnant with a baby that is not at risk of inheriting the HD gene. Couples have to be referred to the specialist clinic by their own genetics clinic, and funding of this procedure can be problematic.

## REGIONAL GENETICS SERVICES

### EAST ANGLIA

**Dept of Clinical Genetics**  
P.O. Box 134  
Addenbrookes Hospital NHS  
Trust  
Cambridge  
CB2 2QQ  
Tel: 01223 216 446

### LONDON-NORTH EAST THAMES

**Clinical Genetics Dept**  
Great Ormond Street Hospital  
London  
WC1N 3JH  
Tel: 0207 762 6845

**Dept of Clinical Neurology**  
The National Hospital for  
Neurology and Neurosurgery  
Queen Square  
London  
WC1N 3BG  
Tel: 0203 448 3613

### LONDON NORTH WEST THAMES

**Kennedy Galton Centre  
for Clinical Genetics**  
Level 8V  
Northwick Park and St Mark's  
NHS Trust  
Watford Road  
Harrow  
Middlesex  
HA1 3UJ  
Tel: 0208 869 2795

### LONDON – SOUTH EAST THAMES

**Dept of Clinical Genetics**  
7<sup>th</sup> Floor  
Guys Hospital  
Great Maze Pond  
London  
SE1 9RT  
Tel: 020 7188 1364

### LONDON – SOUTH WEST THAMES

**Regional Genetics Service**  
St Georges Hospital Medical  
School  
Cranmer Terrace  
London  
SW17 0RE  
Tel: 0208 725 (0571 or 5335)

### MERSEYSIDE

**Mersey Regional Genetic  
Services**  
Royal Liverpool Children's  
Hospital  
Alder Hey  
Eaton Road  
Liverpool  
L12 2AP  
Tel: 0151 802 5002

**Chester Regional Clinical  
Genetics Service**  
The Long House  
Countess of Chester Hospital  
Liverpool Road  
Chester  
CH2 1UL  
Tel: 01244 364 754

### NORTHERN

**Institute of Human Genetics**  
International Centre for Life  
Central Parkway  
Newcastle-upon-Tyne  
NE1 3BZ  
Tel: 0191 241 8721

### NORTH WEST

**Genetic Medicine**  
6<sup>th</sup> Floor St Mary's Hospital  
Oxford Road  
Manchester  
M13 0JH  
Tel: 0161 276 6510

### OXFORD

**Dept of Clinical Genetics**  
The Churchill Hospital  
Old Road  
Headington  
Oxford  
OX3 7LJ  
Tel: 01865 226 024

### SOUTH WEST

**Clinical Genetics Dept**  
St Michael's Hospital  
Southwell Street  
Bristol  
BS2 8EG  
Tel: 0117 342 5115

### Devon and Cornwall Clinical Genetics Service

Heavitree Hospital  
(Royal Devon and Exeter  
Hospital)  
Gladstone Road  
Exeter  
EX1 2ED  
Tel: 01392 405 728

### TRENT

**Leicestershire Genetics Service**  
Leicester Royal Infirmary  
Leicester  
LE1 5WW  
Tel: 0116 258 5736

### Dept of Clinical Genetics

Nottingham University Hospitals  
NHS Trust, City Hospital Campus  
The Gables Gate 3  
Hucknall Road  
Nottingham  
NG5 1PB  
Tel: 0115 962 7728

### Sheffield Clinical Genetic Service

Sheffield Children's Hospital  
Western Bank  
Sheffield S10 2TH  
Tel: 0114 271 7025

**WESSEX  
Wessex Clinical Genetics  
Service**

Princess Anne Hospital  
Coxford Road  
Southampton  
SO16 5YA  
Tel: 02381 206 170

**West Scotland Regional  
Genetics Service**

Level 2, Laboratory Medicine  
Southern General Hospital  
Govan Road  
Glasgow  
G51 4TF  
Tel: 0141 354 9200/9300

**WEST MIDLANDS  
West Midlands Regional  
Clinical Genetics Unit**

Birmingham Women's Hospital  
Mendlesham Way  
Edgbaston, Birmingham  
B15 2TG  
Tel: 0121 627 2630

**Inverness Cytogenetics  
Laboratory**

Raigmore Hospital  
Old Perth Road  
Inverness  
Scotland  
IV2 3UJ  
Tel: 01463 704 000

**YORKSHIRE  
Dept of Clinical Genetics**

Ward 10, Chapel Allerton  
Hospital  
Chapeltown Road  
Leeds  
LS7 4SA  
Tel: 0113 392 4463

**Genetics Unit**

Level 6  
Ninewells Hospital & Medical  
School  
Dundee  
Scotland  
DD1 9SY  
Tel: 01382 632 151

**NORTHERN IRELAND  
Dept of Medical Genetics**

Belfast City Hospital  
Lisburn Road  
Belfast  
BT9 7AB  
Tel: 028 9063 2716

**WALES  
Institute of Medical Genetics**

University Hospital of Wales  
Heath Park  
Cardiff  
CF4 4XW  
Tel: 02920 744 023

**SCOTLAND  
Dept of Medical Genetics**

Medical School  
Ashgrove House  
Foresterhill  
Aberdeen  
AB25 2ZA  
Tel: 01224 552 120

**EIRE  
National Centre for Medical  
Genetics**

Our Lady's Children's  
Hospital  
Crumlin  
Dublin 12  
Ireland  
Tel: 00 353 1409 6902

**Dept of Clinical Genetics**

Western General Hospital  
David Brock Building  
Edinburgh  
EH4 2XU  
Tel: 0131 537 1116

## **Fact sheets available from the Huntington's Disease Association:**

- General information about Huntington's Disease and the Huntington's Disease Association
- Predictive Testing for Huntington's Disease
- Talking to Children about Huntington's Disease
- Information for Teenagers
- Eating and Swallowing Difficulties
- Huntington's Disease and Diet
- The Importance of Dental Care
- Communication Skills
- Behavioural Problems
- Sexual Problems
- Huntington's Disease and the Law
- Huntington's Disease and Driving
- Advice on Life Assurance, Pensions, Mortgages etc.
- Seating Equipment and Adaptations
- Checklist for Choosing a Care Home
- Advance Decision to Refuse Treatment (ADRT)
- A Carers Guide
- A Young Adults Guide
- Challenging Behaviour in Juvenile Huntington's Disease
- A Brief Guide to Juvenile Huntington's Disease for Children's Hospices and Palliative Care Service
- A Young Person with Juvenile Huntington's Disease at School

All fact sheets can be downloaded free of charge from our website [www.hda.org.uk](http://www.hda.org.uk) or ordered from Head Office.

For a publication price list/order form, membership form, details of our Specialist HD Advisers and local Branches and Groups, please call Head Office on:

Tel: 0151 331 5444 or Email: [info@hda.org.uk](mailto:info@hda.org.uk)

Huntington's Disease Association  
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