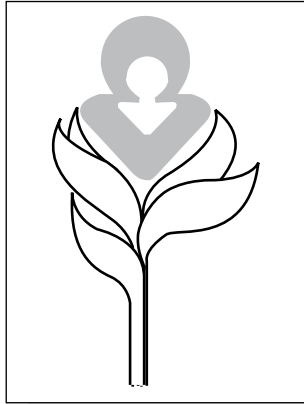


Huntington's Disease Association



Predictive Testing for Huntington's Disease

Fact Sheet

Predictive Testing for Huntington's Disease

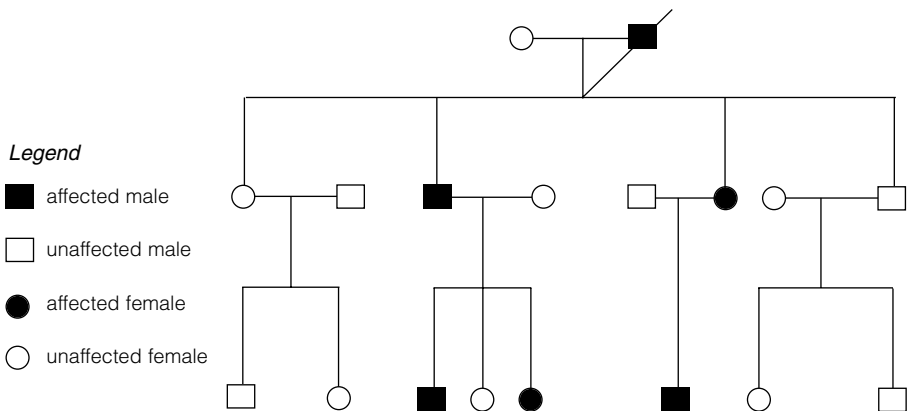
Huntington's disease is an hereditary disorder of the central nervous system. It is caused by a faulty gene. If one of your parents or another relative has Huntington's disease then you will probably be wondering about the likelihood of developing the disease yourself.

This fact sheet explains:

- how Huntington's disease is passed on
- the likelihood of developing the disease
- genetic testing
- the Huntington's gene
- having children – including fetal 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 are advised to sort out any life insurance before being tested. It is probable that a positive test result would make it 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 Genetic 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 Genetic 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 are not usually allowed 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 Genetic Clinic well before embarking on the pregnancy.

Preimplantation Genetic Diagnosis (PGD) offers another alternative to testing for HD in a pregnancy (prenatal testing) and at present it is available to those couples who have received a positive presymptomatic HD result. PGD gives a couple the chance of conceiving a pregnancy that should be unaffected by HD. PGD involves the couple under going 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.

REGIONAL GENETICS SERVICES

EAST ANGLIA

Dept of Clinical Genetics

P.O. Box 134
Addenbrookes Hospital NHS Trust
Cambridge
CB2 2QQ
Tel: 01223 216446

LONDON – NORTH EAST THAMES

Clinical Genetics Unit

Regional Genetics Service
Institute of Child Health
30 Guildford Street
London
WC1N 1EH
Tel: 0207 905 2647

Dept of Clinical Neurology

The National Hospital for
Neurology and Neurosurgery
Queen Square
London
WC1N 3BG
Tel: 0207 837 3611

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

Division of Medical & Molecular Genetics

8th Floor
Guy's Tower
Guys Hospital
St Thomas Street
London
SE1 9RT
Tel: 0207 7188 1364

LONDON – SOUTH WEST THAMES

Regional Genetics Service

St Georges Hospital Medical
School
Cranmer Terrace
London
SW17 0RE
Tel: 0208 8725 5335

MERSEYSIDE

Mersey Regional Genetic Services

Royal Liverpool Children's
Hospital
Alder Hey
Eaton Road
Liverpool
L12 2AP
Tel: 0151 252 5238

Chester Regional Clinical Genetics Service

Moston Lodge
Countess of Chester Hospital
Liverpool Road
Chester
CH2 1UL
Tel: 01244 364754

NORTHERN

Institute of Human Genetics

International Centre for Life
Central Parkway
Newcastle-upon-Tyne
NE1 3BZ
Tel: 0191 241 8721

NORTH WESTERN

Dept of Medical Genetics

St Mary's Hospital
Hathersage Road
Manchester
M13 0JH
Tel: 0161 276 6002

Manchester Paediatric Genetics Unit

Royal Manchester Children's
Hospital
Hospital Road
Pendlebury
Manchester
M27 4HA
Tel: 0161 727 2335

OXFORD

Dept of Clinical Genetics

The Churchill Hospital
Old Road
Headington
Oxford
OX3 7LJ
Tel: 01865 226066

SOUTH WESTERN

Clinical Genetics Service

St Michael's Hospital
Southwell Street
Bristol
BS2 8EG
Tel: 0117 9285652

**Devon and Cornwall Clinical
Department of Genetics**

Dept of Child Health

Royal Devon and Exeter
Hospital (Heavitree)
Gladstone Road
Exeter
EX1 2ED
Tel: 01392 405726

TRENT

Leicestershire Clinical Genetics

Leicester Royal Infirmary
Leicester
LE1 5WW
Tel: 0116 258 5736

Dept of Clinical Genetics

2nd Floor
H Block
City Hospital NHS Trust
Hucknall Road
Nottingham
NG5 1PB
Tel: 0115 962 7728

**North Trent Clinical Genetic
Service**

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

WESSEX

**Wessex Clinical Genetics
Service**

Princess Anne Hospital
Coxford Road
Southampton
SO16 5YA
Tel: 02380 796166

WEST MIDLANDS

**West Midlands Clinical
Genetics Unit**

Birmingham Women's Hospital
Metchley Park Road
Edgbaston
Birmingham
B15 2TG
Tel: 0121 627 2630

YORKSHIRE

Dept of Clinical Genetics

Ashley Wing
St James University Hospital
Beckett Street
Leeds LS9 7TF
Tel: 0113 206 5555

NORTHERN IRELAND

Dept of Medical Genetics

Floor A Belfast City Hospital
Lisburn Road
Belfast
BT9 7AB
Tel: 028 9026 3873

SCOTLAND

Department of Medical Genetics

Medical School
Foresterhill
Aberdeen AB25 2ZD
Tel: 01224 552 120

Dept of Clinical Genetics

Western General Hospital
Crewe Road
Edinburgh EH4 2XU
Tel: 0131 651 1012

West Scotland Regional Genetics Service

Ferguson Smith Centre for
Clinical Genetics
Block 4 Yorhill NHS Trust
Glasgow G3 8SJ
Tel: 0141 201 0808

Inverness Cytogenetics Laboratory

Raigmore Hospital
Old Perth Road
Inverness
Scotland IV2 3UJ
Tel: 01463 704000

Department of Human Genetics

Level 6
Ninewells Hospital & Medical
School
Dundee
Scotland
DD1 9SY
Tel: 01382 632035

WALES

Institute of Medical Genetics

University Hospital of Wales
Heath Park
Cardiff CF4 4XW
Tel: 02920 744028

EIRE

National Centre for Medical Genetics

Our Lady's Children's Hospital
Crumlin
Dublin 12
Ireland
Tel: 00 353 1409 6739

Fact sheets available from the HDA:

- General Information about Huntington's Disease and the HDA
- 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 Carer's 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 for free from our website:
www.hda.org.uk or ordered direct from Head Office

For a publication price list/order form, membership form, details of our Regional Care Advisers and local Branches and Groups, please telephone, email or write to:

Huntington's Disease Association

Neurosupport Centre, Norton Street, Liverpool L3 8LR

Tel: +44 (0)151 298 3298 Fax: +44 (0)151 298 9440

Email: info@hda.org.uk Web: www.hda.org.uk