

# Huntington disease and genetic testing





First published 1997  
This edition 2010



**This information is for people who:**

- **have been diagnosed with Huntington disease**
- **have a history of Huntington disease in their family.**

**The booklet provides information about:**

● The family condition called Huntington disease (HD) Pg1

● Genetic counselling and testing for Huntington disease Pg12

● Frequently asked questions about genetic testing Pg18

● Factors for you to consider about genetic testing Pg23

● Research and Huntington disease Pg26

● Further information and support Pg27



## The family condition called Huntington disease (HD)

### What is HD?

HD is an inherited neurological condition affecting the brain and nervous system. It is a genetic condition that causes involuntary muscle movements, personality changes, and reduced memory and reasoning abilities.

Symptoms commonly associated with HD, previously known as Huntington's chorea, were recorded in European populations in the 1600s. The condition is named after Dr George Huntington who described the inheritance of HD as early as 1872.

### How does HD affect people?

The first noticeable symptoms of HD are usually muscle twitches and/or changes in personality. Symptoms commonly begin to appear in men or women between the ages of 30 and 50, although HD can appear at any age.

In some families the age when symptoms first appear (age of onset) and the effects of HD might be similar. However, these might also differ considerably among members of the same family.

Symptoms of HD vary and can include:

- involuntary muscle movements affecting the face, hands, limbs and body that are irregular, jerky and uncoordinated
- slurred speech and difficulties with swallowing
- disruptions to thought processes, including short term memory loss, difficulty with making decisions, lack of motivation, reduced insight and awareness of social boundaries
- unsteadiness and falls
- difficulty with emotional control, frustration, agitation and mood swings
- depression and anxiety.

HD progresses slowly. After developing the first symptoms, affected people usually live 15 to 20 years.

## How common is HD?

About 1 in 15,000 Australians is affected by HD. It affects males and females equally.

The condition is found in all ethnic groups but is more common among those of European descent.

## What is the treatment?

Currently, there is no definitive treatment or cure for HD. However, research has led to improvements in the management of symptoms.

Medical researchers continue to investigate the effects of HD on the brain in the hope of finding effective treatments to delay the onset of the condition and to slow its progress.

## What causes HD?

HD is caused by a change in the genetic information that is passed on from parents to children and contributes to growth, development and health. Genetic information is in the form of many thousands of different genes.

Changes often occur in genes, but most do not cause health problems. HD is caused by a change in a gene that makes the gene faulty and stops it from working properly. This is called a gene mutation.

The gene associated with HD is known by a variety of names - Htt, IT15, and HD gene. The term huntingtin gene will be used in this booklet.

Huntingtin genes tell brain cells to make a protein called huntingtin which plays an important role in brain function. A faulty huntingtin gene produces a different form of huntingtin protein which builds up in parts of the brain and is associated with HD later in life.

**HD is due to an inherited change in a huntingtin gene.**

## Why does HD occur in several generations of some families?

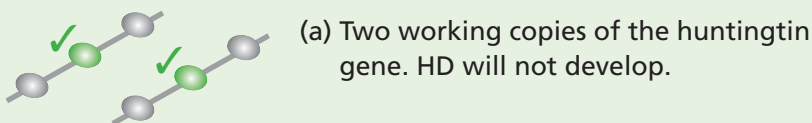
HD is called a **dominant genetic condition**. When HD occurs in a family, it often appears in one or more members in each generation.

Everyone is born with two copies of the huntingtin gene in every cell of their body. People affected by HD have inherited a **faulty** huntingtin gene copy and a working huntingtin gene copy (Figure 1a).

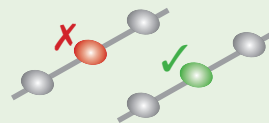
A faulty huntingtin gene copy dominates over a working copy and HD will almost certainly develop if the person lives a normal life expectancy (Figure 1b).

A person who has inherited a faulty huntingtin gene copy but is not yet affected by HD is said to be **presymptomatic** for HD.



Figure 1. Impact of a faulty huntingtin gene copy.



(b) One faulty copy and one working copy. HD will almost certainly develop if the person lives a normal life expectancy.



**KEY**

-  Faulty copy of the huntingtin gene
-  Working copy of the huntingtin gene

## How is HD passed on in families?

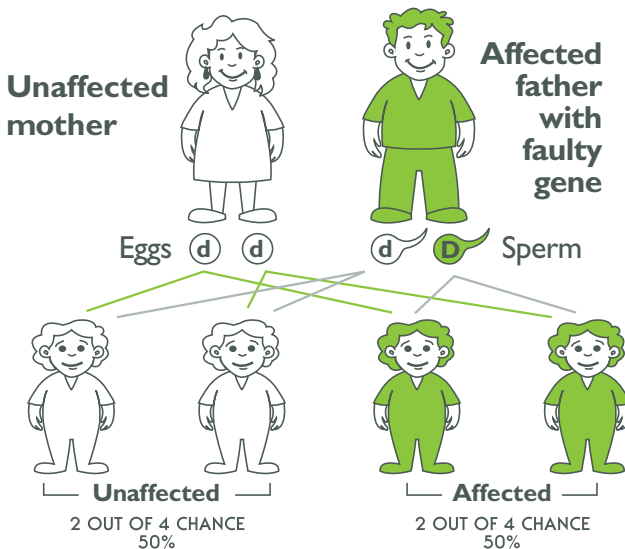
Each parent passes one of their two huntingtin gene copies on to their child.

A person affected by, or presymptomatic for, HD was born with a faulty huntingtin gene copy.

When that person becomes a parent, as shown in Figure 2, there is a 50% chance that they will pass on the faulty gene copy to their child.

In the example below (Figure 2), the father who is affected by or presymptomatic for HD has a faulty huntingtin gene copy 'D'. He also has a working huntingtin gene copy 'd'. However, the parent could be either the mother or the father.

Figure 2. Inheritance pattern when one parent has a faulty huntingtin gene copy



As shown in Figure 2, in every pregnancy, parents pass on to their children one of four possible combinations of huntingtin gene copies to their children:

- There is a 2 out of 4 chance (50%) that a child will inherit the two working huntingtin gene copies 'dd'  
The child will not be affected by HD.
- There is a 2 out of 4 chance (50%) that a child will inherit a faulty gene copy and a working copy.  
The child will almost certainly be affected by HD if they live a normal life expectancy.

A 50% chance does not mean that exactly half the children of an affected or presymptomatic parent will inherit the faulty gene copy. Nor does it mean that every second child in the family will inherit it. The 50% chance applies to each pregnancy.

In some families HD symptoms might occur at an earlier age with each new generation. This is more likely to happen if the affected or presymptomatic parent is the father than the mother.

**HD is a condition that runs in families. Informing other family members of a diagnosis of HD will allow them to be fully informed when making life decisions.**

## The information in the huntingtin gene

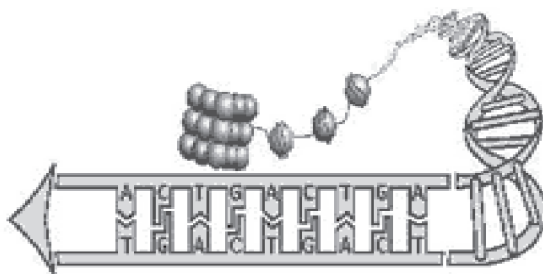
Genes are segments of DNA (deoxyribonucleic acid). DNA is a chemical that produces a coded message for cells to make proteins and other products.

As shown in Figure 3, the code is made up of the letters A,C,G and T. A gene is a long sequence of thousands of these letters.

Each 'word' in the coded message is made up of three of these letters e.g. CAG or TCA. Code words are referred to as 'triplets'.

A small segment of the huntingtin gene copies includes a CAG triplet which is repeated many times.

Figure 3. The letters A, C, G and T make up the information in the DNA



## The triplet repeat in the huntingtin gene

Huntingtin gene copies with 26 or less CAG triplet repeats are described as being in the normal range.

...CAG-CAG-CAG...26 or less repeats...

The **normal range** for the number of CAG repeats

## An expansion of the triplet repeat in the huntingtin gene

A change in a huntingtin gene copy which results in it containing more than 26 CAG repeats is described as an **expansion**.

The number of CAG repeats in an expanded gene copy determines if it will work properly.

**When the huntingtin gene copy contains more than 40 CAG repeats, it is described as faulty.**

**HD is caused by a gene copy with an expansion of 40 or more CAG repeats.**

For a small number of people an expansion of 36-39 CAG repeats is also likely to cause HD.

You will find a summary of current knowledge of the impact of the ranges of CAG repeats in the huntingtin gene and what it means for you and your children on pg 9 and 10.

## What the number of repeats in an expanded huntingtin gene copy means for you and your children

Number of CAG repeats*	What does it mean for you?
26 or less Normal range	You will not develop HD.
27 – 35 Intermediate range	You will not develop HD.
36 – 39 Increased risk range (reduced penetrance range)	You are likely to develop HD in your lifetime. However: <ul style="list-style-type: none"><li>• you might develop it at a late age</li><li>• the condition might be less severe</li><li>• you might not develop HD at all.</li></ul>
40 or more faulty gene HD range (full penetrance range)	You will almost certainly develop HD if you live a normal life expectancy.

\*The ranges of CAG repeats are in accordance with Laboratory Guidelines for Huntington Disease Genetic Testing (1998) produced by the American College of Medical Genetics/ American Society of Human Genetics Huntington Disease Genetic Testing Working Group.

## What does it mean for your children?

Your children will not develop HD.

Each child has a 50% (1 in 2) chance of inheriting your expanded huntingtin gene copy with 27-35 CAG repeats. If they do, they are not likely to develop HD.

However, there is a small chance that the gene will have expanded further than 35 repeats. In this case, your child is likely to develop HD in their lifetime. This is more likely to happen through the father than the mother.

Each child has a 50% (1 in 2) chance of inheriting your expanded huntingtin gene copy with 36–39 CAG repeats. If they do, they are likely to develop HD in their lifetime. However:

- they might develop it at a late age
- the condition might be less severe
- they might not develop HD at all.

There is a chance that the gene will have expanded further than 39 repeats. This is more likely to happen through the father than the mother. In this case, see below.

Each child has a 50% (1 in 2) chance of inheriting your faulty huntingtin gene copy with 40 or more CAG repeats. If they do they will almost certainly develop HD if they live a normal life expectancy.

### **What if HD is not known to have affected other family members?**

A small number of people with HD do not have a known family history of the condition at the time of testing.

This can happen if one of the unaffected parents passes on a huntingtin gene copy which expands and becomes a faulty gene copy in the next generation. The expansion is more likely to come through a father than a mother.

The size of an expansion in the huntingtin gene impacts on the effects of HD in following generations. See Summary pages 9 and 10.

**In this case, genetic test results for the unaffected parents could provide important information for their family.**

## Genetic counselling and testing for Huntington disease

### A clinical diagnosis

If it is thought that you are already experiencing symptoms of HD your family doctor will usually provide a referral to a neurologist. A neurologist will conduct a comprehensive assessment and provide a clinical diagnosis and treatment plan.

### The diagnostic genetic test

Following a clinical diagnosis a neurologist might advise you to consider a diagnostic genetic test. A genetic test result, especially if you have no known family history of HD, could support a clinical diagnosis and avoid unnecessary testing for other conditions with similar symptoms to HD.

### Genetic counselling

If you are affected by HD or have a family history of HD, your doctor might suggest an appointment with your nearest genetic counselling service or HD clinic. Contact details are on pg 27.

A genetic counsellor will discuss the implications of a diagnosis of HD for your family. A genetic counselling team will provide support for you and your family while you are making decisions about genetic testing or prenatal testing options.

### The presymptomatic genetic test

If you have a blood relative affected by HD, but do not have symptoms of HD yourself, you are said to be 'at risk' for developing the condition.

You might wish to have a **presymptomatic genetic test** to find out if you have inherited an expanded huntingtin gene copy. The genetic test for HD investigates the **number of CAG triplet repeats** in a huntingtin gene.

Presymptomatic testing, also known as predictive testing, is the term used in this booklet. Presymptomatic testing is used because every person whose genetic test result reveals a faulty gene copy will almost certainly develop symptoms if they live a normal life expectancy.

The presymptomatic genetic test for HD cannot determine when symptoms will begin or how severe symptoms are likely to be. If a faulty huntingtin gene copy is detected, you might remain healthy for some years before symptoms begin.

If a genetic test shows that you have inherited a faulty huntingtin gene copy, you could request a neurological assessment. The assessment can determine if symptoms of HD are present and provide a professional contact person for future care.

### Who can have the presymptomatic genetic test?

According to international guidelines if you are 18 years or older and have a blood relative who has been diagnosed with HD, you are eligible for the genetic test.

In accordance with NSW Health policy, presymptomatic genetic testing for HD can only be accessed through a genetic counselling service.

## The presymptomatic genetic testing process

The testing process usually includes:

- An initial appointment with a genetic counselling team to record your family history of HD, provide information about the inheritance of the condition and discuss the option of genetic testing.
- A blood sample will be required if you decide to have a genetic test - either at the appointment or another time that suits you.
- Another appointment within a few months to receive the test results (if you still wish to receive the results).
- Follow up counselling and support.

## Genetic test results

For more information about possible genetic test results, see pg9 and 10. Follow up counselling and support is provided regardless of the result.



### Genetic testing and reproductive choices

There are various options available to you if you do not want to risk passing an expanded huntingtin gene copy on to a child.

Reproductive choices include **prenatal genetic testing** and **pre-implantation genetic diagnosis (PGD)**.

**It is recommended that you discuss prenatal testing for HD with a genetic counselling team before pregnancy.**

### Prenatal genetic testing

You might request prenatal testing if you or your partner have a family history of HD or have been clinically diagnosed with HD.

The purpose of prenatal genetic testing is to determine whether your unborn baby has inherited an expanded huntingtin gene copy.

It is important to note that if you or your partner have a family history of HD but do not have symptoms, a prenatal result which shows that the baby has an expanded huntingtin gene copy will automatically reveal an expanded gene copy in a parent. Finding out that both a parent and a child could develop HD might be difficult to accept.

If there is time, it is preferable that the parent be tested first followed by prenatal testing.

### Prenatal genetic testing options

#### **Chorionic villus sampling (CVS)**

CVS is usually carried out between 11 and 12 weeks of pregnancy. Therefore, it is advisable to see a doctor as soon as a pregnancy begins.

CVS involves taking a small sample of cells from the chorion, which is the lining of the uterus that later develops into the placenta. Under the guidance of ultrasound, a small sample of chorion is removed for testing by passing a fine needle through the cervix or through the abdominal wall. A local anaesthetic is usually used prior to the procedure.

The sample is tested for an expanded huntingtin gene copy. Laboratories aim to provide a result within a short time frame.

When CVS is carried out by an obstetrician who is experienced in the technique the risk of miscarriage related to the test is less than 1% (less than 1 in 100 pregnancies).

#### **Amniocentesis**

Amniocentesis is usually carried out between 15 and 19 weeks of pregnancy. Under the guidance of ultrasound, a small sample of amniotic fluid is removed for testing by passing a fine needle through the abdominal wall and into the amniotic fluid surrounding the baby. The amniotic fluid contains cells of the developing baby. A local anaesthetic is usually used prior to the procedure.

The sample is tested for an expanded huntingtin gene copy. Laboratories aim to provide a result within a short time frame.

When amniocentesis is carried out by an obstetrician who is experienced in this technique, the risk of miscarriage related to the test is less than 1% (less than 1 in 100 pregnancies).

### **Pre-implantation genetic diagnosis (PGD)**

As an alternative to prenatal testing, some couples choose in-vitro fertilisation (IVF) to create embryos that are tested for an expanded huntingtin gene. An unaffected embryo is implanted into the mother's uterus.

One of the benefits of PGD is that decisions regarding pregnancy termination can be avoided. Currently, PGD is only available through private fertility clinics.

### **How can I find out more about prenatal testing or PGD?**

Genetic counselling services or HD clinics can provide you with more information. Contact details are on pg 27.

Prenatal testing information is also available from the Centre for Genetics Education [www.genetics.edu.au](http://www.genetics.edu.au).

## Frequently asked questions about genetic testing

### **Do I have to have a genetic test?**

No. A person with a family history of HD is not obliged to find out if they have inherited an expanded huntingtin gene copy.

Deciding to seek genetic counselling does not mean that you have to have a genetic test. After obtaining information many people choose not to have the genetic test.

### **How accurate is the test?**

The genetic test result is near 100% accurate.

### **Will everyone get a definite answer?**

Almost everyone undergoing the test will receive a definite result.

For the small number of people whose test result shows an expansion in the reduced penetrance range (36-39 repeats), it is not possible to predict with certainty if they will develop HD.

### **Can the result indicate my age when HD symptoms will start?**

In general, the larger the number of CAG repeats the earlier the age of onset of HD, but it is not possible to predict the exact age of onset.

Other factors, such as underlying health conditions, environment, lifestyle, or other genes might also influence the age of onset. Even though people might have the same CAG repeat number, there can be a large variation in the age that symptoms start.

### **Is there anything I can do to delay the start of HD?**

There is some evidence that a healthy lifestyle, keeping mentally and physically fit, and reducing or avoiding things that could adversely affect brain health (e.g. alcohol and high blood pressure) could help you delay the onset of HD.

### **How long will the presymptomatic genetic test take?**

The process usually takes approximately three months depending on individual circumstances, laboratory workload and the resources available.

You can choose if and when you would like to receive the results. You can delay receiving a result if there are life circumstances that make it an inappropriate time to receive a result.

### **If I have suspected symptoms of HD but no known family history can I have a genetic test?**

Yes. A genetic test will detect the presence of an expanded huntingtin gene copy whether or not you have a known family history of HD.

If you have symptoms similar to HD and the genetic test does not detect an expanded huntingtin gene copy, this might indicate the presence of another neurological condition.

### **If I have been diagnosed with HD, do my blood relatives need to have the genetic test?**

It is important that your blood relatives know that they are at risk for developing HD and that a genetic test is available.

Some family members might wish to assist by having the genetic test but might not want to find out their personal test result.

Others might decide to have their DNA stored for a possible future test. This is called DNA banking. Information about DNA banking is available from your genetic counselling or HD service. Contact details are on pg 27.

### **Can other people find out about my test results?**

In accordance with the *Health Records and Information Privacy Act 2000* your result will not be given to anyone without your written consent.

An exception to this might be that the information is required by a court of law.

### **What is the cost of genetic testing?**

You will not be charged for genetic testing when it is provided through a NSW genetic counselling service or HD clinic.

### How might my test result affect my insurance policies?

#### Health insurance:

You are not required to disclose your family history of HD or your genetic test result to private health insurance companies.

#### Life insurance including trauma/disability and income protection policies.

- **Applying for a new policy.** You are required to tell the insurer about any genetic information that you know at the time of taking out a new policy. This includes information about your health and your family history of HD, and the result of a genetic test if you or your relatives have had one. Most companies ask specifically about family health history and genetic testing.
- **An existing life insurance policy.** If you have a genetic test which shows that you have inherited the expanded or faulty huntingtin gene copy, your results should not affect an existing insurance policy. You do not have to tell the insurance company of test results and your policy will stay in place as long as payments are made.
- **Changing your existing policy.** You will be required to tell the insurance company your genetic test result if you change a policy. You will also be required to tell them any new information you know about the health of family members, including genetic test results for HD.

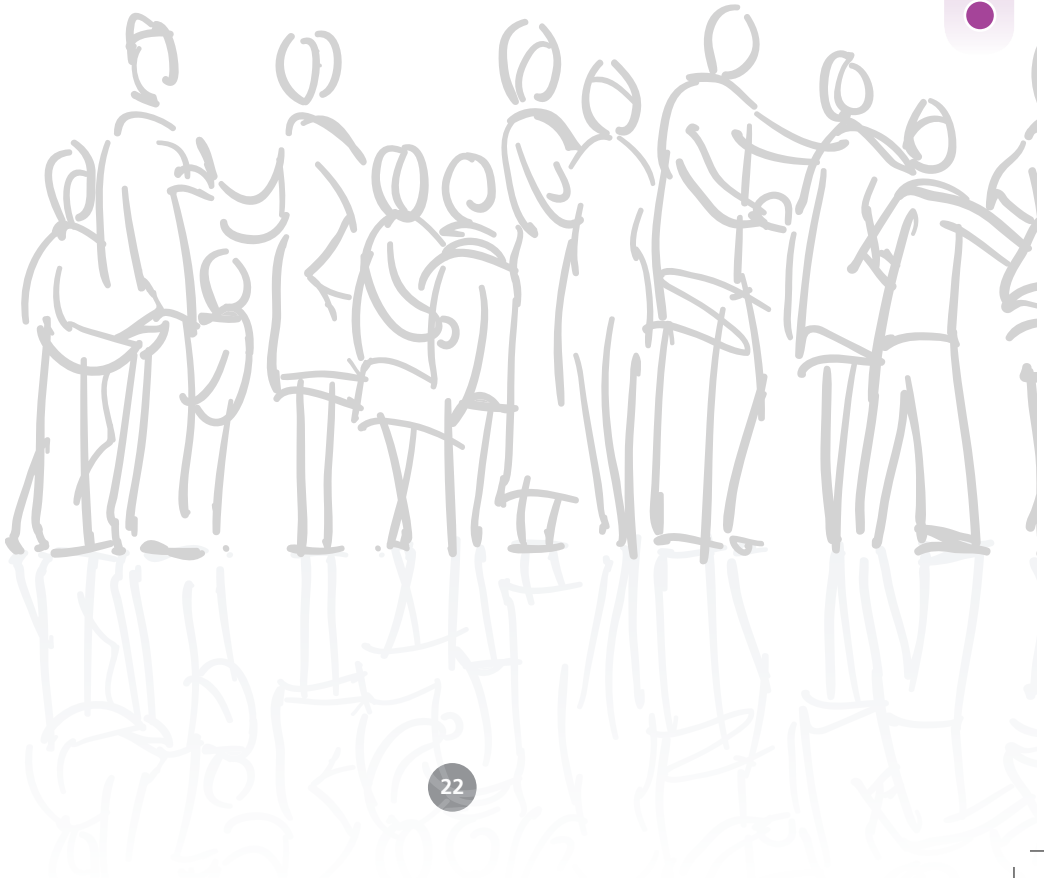
If your insurance company is aware of HD in your family and a future result shows that you have not inherited the expanded or faulty huntingtin gene copy, you should tell the insurer and ask to have your premium re-assessed.

The Investment and Financial Services Association (IFSA) is an organisation representing most of the insurance companies in Australia. They have agreed that its insurance companies will:

- not require someone to have a genetic test
- not use family members' results to assess other family members' policies when applying to the same company
- ensure confidentiality of your genetic test result.

IFSA ph (02) 9299 3022

More information is available from the Centre for Genetics Education ph (02) 9926 7324 or [www.genetics.edu.au](http://www.genetics.edu.au).



## Factors for you to consider about genetic testing

### **Different people, different feelings**

There is no right or wrong way to feel about having a genetic test for HD. Life circumstances are different for everyone.

Family or friends might also have ideas about whether or not you should have the test. Sometimes it can be difficult for other family members to accept your decision. Each person has the right to make their own decision.

It might be helpful for you to see a genetic counsellor for information and support.

If you decide to have the test, it could be difficult to try to go through the testing process alone. You might consider asking a friend or family member to accompany you to appointments.

Genetic testing might also have an impact on those close to you. In particular, your partner might find the process difficult. They could feel left out or anxious because they are not blood relatives and yet the results will be important for family planning and could possibly impact on your life together.

It is important to think about the effect of genetic testing on your extended family. Sometimes family members who receive the same result will feel closer to each other. Family members who receive different results might feel uncomfortable with each other and find it harder to talk to each other.

It might be helpful for you to think about any potential differences of opinion in your family ahead of time. You might like to consider the best ways to handle this before deciding about a genetic test. These issues could also be discussed with a genetic counsellor.

### Weighing up the pros and cons of genetic testing

The process of deciding whether or not to have a genetic test might be easy for some people and difficult for others. There are many questions to consider:

- Why is a test result important to me?
- How could the result affect my future?
- What will I tell my children?
- How do I feel about having children and prenatal testing?
- How could the result (positive or negative) affect my role in my family?

A test result that shows that you will not develop HD can also cause conflicting thoughts and feelings. As HD is a condition that runs in families, this test result might mean that you could become a carer within the family.

You might find it helpful to make a record of pros and cons (positives and negatives) at the time of decision making.

## Factors to consider about testing

Here is a list of pros and cons from other peoples' experiences. You might have your own list for your specific family situation.

**Table 2. What others have said about the pros and cons of genetic testing**

Some pros	Some cons
<i>"It will help for planning a family."</i>	<i>"I prefer to live my life with hope."</i>
<i>"We won't have to live with uncertainty if I have a test result."</i>	<i>"We have always known about HD in our family and are happy with not having testing. We don't need to know."</i>
<i>"If I find out I don't have the faulty gene, I won't have the burden of worrying if I will get HD and I can plan my future better."</i>	<i>"I will have to think about how the test result might affect my family ... and my job."</i>
<i>"The test result will help my sister and her family."</i>	<i>"If the test shows the faulty gene, it could be a long time before there is a cure ..."</i>
<i>"If I find out I do have the faulty gene I might be able to join some treatment or research trials."</i>	<i>"I have coped with other difficult times OK."</i>
<i>"We would like to stop HD in our family."</i>	<i>"My test result might make my children worried."</i>

## Research and Huntington Disease

A widespread international effort is underway to find a cure for HD or ways of slowing the progression and severity of symptoms.

The major international research programs are the Huntington Study Group (HSG) and the European HD Network.

The HSG was formed by a group of researchers in 1993. It combines research faculties with teams of professionals who have expertise in treating HD. The group has begun to test new drugs that could lead to effective treatments.

The European HD Network is a large group of medical and scientific professionals across Europe working with people and families affected by HD. The Network undertakes studies and large scale clinical drug trials.

Australian researchers are working with these groups as well as undertaking independent research, including drug trials.



## Further information and support

Centres that offer genetic counselling services in NSW are listed below. Community support is also available for people with HD. Your doctor will inform you about what is available in your area.

**The Huntington Disease Service**, Sydney West Area Health Service, is based at The Children's Hospital, Westmead. The Service consists of an HD specific clinic (ph (02) 9845 6544), research centre (ph (02) 9845 6793), genetic counselling (ph (02) 9845 6544), multidisciplinary assessment (ph (02) 9804 5863) and Huntington Lodge which provides inpatient and respite care (ph (02) 9845 5803).

### 1. Genetic Counselling services and HD Clinics for NSW & ACT

Interpreter services are available.

#### Metropolitan areas

##### CAMPERDOWN

Department of Molecular and Clinical Genetics  
Royal Prince Alfred Hospital  
Ph (02) 9515 5080

##### KOGARAH

Clinical Genetics Service  
St George Hospital  
Ph (02) 9113 3635

##### LIVERPOOL

Clinical Genetics Department  
Liverpool Hospital  
Ph (02) 9828 4665

##### NEWCASTLE

Huntington Disease Clinic  
Hunter Genetics  
Waratah  
Ph (02) 4985 3100

##### PENRITH

Clinical Genetics Department  
Nepean Hospital  
Ph (02) 4734 3362

##### RANDWICK

Department of Medical Genetics  
Sydney Children's Hospital  
Ph (02) 9382 1704

##### ST LEONARDS

Department of Clinical Genetics  
Royal North Shore Hospital  
Ph (02) 9926 6478

##### WESTMEAD

Department of Clinical Genetics  
The Children's Hospital  
Westmead  
Ph (02) 9845 3273

##### WOLLONGONG

Block C, Level 2 Antenatal Clinic  
Wollongong Hospital  
Crown Street  
Ph (02) 4253 4267

##### CANBERRA

ACT Genetics Service  
The Canberra Hospital  
Ph (02) 6244 2133

### NSW Outreach Services

#### BATHURST

Bathurst Community Health Centre  
Level 3, Bathurst Base Hospital  
Howick Street  
Ph (02) 6330 5677

#### COFFS HARBOUR

Coffs Harbour Health Campus  
345 Pacific Highway  
Ph (02) 6656 7200

#### DUBBO

Dubbo Community Health Centre  
2 Palmer Street  
Ph (02) 6885 8937

#### FORSTER

Forster Community Health Centre  
Breeze Parade  
Ph (02) 6555 1800

#### GOSFORD

Child and Family Health Centre  
Level 1, Gateway Centre  
237 Mann Street  
Ph (02) 4328 7994

#### GOULBURN

Child, Infant and Family Tertiary Service  
Faithful Street  
Ph (02) 4827 3950

#### KINGSCLIFF

Kingscliff Community Health Centre  
Turnock Street  
Ph (02) 6674 9500

#### LISMORE

Lismore Community Health Service  
29 Molesworth Street  
Ph (02) 6620 2967

#### PORT MACQUARIE

Port Macquarie Community  
Health Centre  
Morton Street  
Ph (02) 6588 2882

#### TAMWORTH

Community Health Centre  
Johnson House  
Tamworth Hospital  
Dean Street  
Ph (02) 6767 8100

#### TAREE

Taree Community Health Centre  
Pulteney Street  
Ph (02) 6592 9703

#### WAGGA WAGGA

Community Health Centre  
Docker Street  
Ph (02) 6938 6443

**Contacts for genetic counselling services outside of NSW can be found at [www.genetics.edu.au](http://www.genetics.edu.au).**

## 2. Support Organisations

### NSW

#### **Australian Huntington's Disease Association (NSW) Inc**

The HD Association provides support and contacts for people affected by HD and their families.

PO Box 178 WEST RYDE NSW 1685  
Ph (02) 9874 9777 Fax (02) 9874 9177  
[www.ahdansw.asn.au](http://www.ahdansw.asn.au)

Copies of the booklets *Huntington Disease* and *Presymptomatic Testing – The Consumer Experience*, are available from the HD Association.

#### **Association of Genetic Support of Australasia (AGSA)**

AGSA provides information about Support Groups. They also provide support for rare conditions that don't have a specific support group.

66 Albion Street SURRY HILLS NSW 2010  
Ph (02) 9211 1462 Fax (02) 9211 8077  
[www.agsa-geneticsupport.org.au](http://www.agsa-geneticsupport.org.au)

### Other states

#### **Australian Huntington's Disease Association (Qld) Inc**

385 Ipswich Road PO Box 635 ANNERLEY QLD 4103  
Ph (07) 3391 8833 Fax: (07) 3391 0443  
[www.qahda.com](http://www.qahda.com)

#### **Australian Huntington's Disease Association (SA & NT) Inc**

PO Box 580 NORTH ADELAIDE SA 5006  
Ph (08) 8271 2922

#### **Australian Huntington's Disease Association (Tas) Inc**

PO Box 1168 BURNIE TAS 7320  
Ph (03) 6431 3403

#### **Australian Huntington's Disease Association (Vic) Inc**

607 Warrigal Road ASHWOOD VIC 3147  
PO Box 60 Holmesglen VIC 3148  
Ph (03) 9563 3922 Toll free 1800 063 501  
[www.ahda.com.au](http://www.ahda.com.au)

#### **Australian Huntington's Disease Association (WA) Inc**

Centre for Neurological Support  
The Niche, Suite B  
11 Aberdare Road NEDLANDS WA 6009  
Ph (08) 9346 7599 Fax (08) 9346 7597  
[www.wa.ahda.asn.au](http://www.wa.ahda.asn.au)

### 3. Further information and Resources

#### **Centre for Genetics Education NSW Health**

Royal North Shore Hospital  
PO Box 317, St Leonards NSW 1590  
Ph (02) 9926 7324 Fax (02) 9906 7529  
[www.genetics.edu.au](http://www.genetics.edu.au)

The Centre provides information and resources for HD (as well as contact details for genetic counselling services).

### References

<sup>1</sup>Nance MA et al. 1998. ACMG/ASHG STATEMENT Laboratory Guidelines for Huntington Disease Genetic Testing. The American College of Medical Genetics/American Society of Human Genetics Huntington Disease Genetic Testing Working Group. *Am J Hum Genet.* 62 (5): 1243-1247.

Hendricks AE et al. 2009. Estimating the Probability of de novo HD Cases from Transmissions of Expanded Penetrant CAG Alleles in the Huntington Disease Gene from male Carriers of High Normal Alleles [27 -35 CAG]. *Am J Med Genet Part A* 149A(7):1375-1381.

Semaka A, Creighton S, Warby S, Hayden M. 2006. Predictive testing for Huntington disease: interpretation and significance of intermediate alleles. *Clin Genet* 70(4):283-294.

Langbehn et al. 2009. CAG-repeat length and the age of onset in Huntington disease (HD): A review and validation study of statistical approaches. *Am J Med Genet Part B: Neuropsychiatric Genetics.*

Walker FO. 2007. Huntington's Disease. *The Lancet.* Vol 369(9557):218-28.

National Organisation for Rare Disorders (NORD) [online]. Available: [www.rarediseases.org/](http://www.rarediseases.org/) [Accessed March 2009]

Online Mendelian Inheritance in Man, OMIM. [online]. Available: [www.ncbi.nlm.nih.gov/omim](http://www.ncbi.nlm.nih.gov/omim) [Accessed March 2009]

### Edit history

The original series of brochures, *Predictive Testing for Huntington disease*, was produced by Associate Professor Kristine Barlow-Stewart, Director, Centre for Genetics Education, in conjunction with Ms Fiona Richards, Huntington Disease Service, the Children's Hospital, Westmead, in 1997.

The information was revised in 2009 by Lynne Purser in collaboration with Associate Professor Kristine Barlow-Stewart, Kate Dunlop, and Jacinta Reid of the Centre for Genetics Education, NSW Health.

We would like to thank the people who have experienced Huntington disease in their families for generously contributing to the project.

Many thanks are also extended to the following for their expert assistance: Mr John Conaghan, Ms Kim Frumar, Dr Marcus Hinchliffe, Dr Clement Loy, Dr Elizabeth McCusker, Ms Fiona Richards, Mr Dominic Ross, Dr Desiree du Sart and Dr Anne Turner.

### IMPORTANT

The information in this brochure is current at the date of production. Please check with your doctor or local genetic counselling service for any new information.



**The Centre for  
Genetics Education**

PO Box 317  
St Leonards NSW 1590  
Ph: (02) 9926 7324 Fax: (02) 9906 7529  
Email: [contact@genetics.com.au](mailto:contact@genetics.com.au)  
[www.genetics.edu.au](http://www.genetics.edu.au)

*A state wide service of the NSW Genetics Service which aims to raise the awareness of the contribution of genetics to family health, both for health professionals and the public. The Centre can be contacted regarding availability of and access to a range of genetic services, information and resources. Referral to metropolitan and outreach genetic clinics and genetic support groups is available.*

The Centre for Genetics Education  
2010

**Further copies of this booklet are available from:**

**The Centre for Genetics Education**  
PO Box 317  
St Leonards NSW 1590  
P: (02) 9926 7324 FAX: (02) 9906 7529  
Email: [contact@genetics.com.au](mailto:contact@genetics.com.au)  
[www.genetics.edu.au](http://www.genetics.edu.au)