

The early symptoms include:

- Slight, uncontrollable muscular movements.
- Stumbling and clumsiness.
- Lack of concentration.
- Short-term memory lapses.
- Depression.
- Changes of mood, sometimes including aggressive or antisocial behaviour.

How HD progresses

Later on in the illness many people experience symptoms which may include:

- Involuntary movements.
- Difficulty in speech and swallowing.
- Weight loss.

As well as emotional changes resulting in:

- Stubbornness.
- Frustration.
- Mood swings.
- Depression.

These symptoms can result in a loss of drive, initiative and organisational skills, which may result in the person appearing to be lazy. There also may be difficulty in concentrating on more than one activity at a time.

Sometimes, psychological problems, rather than the physical deterioration, cause more difficulties for both the person with HD and their carers. Some changes are definitely part of the disease process although they may be made worse by other factors. It is depressing to have a serious illness and extremely frustrating not to be able to do things which previously seemed simple.

In the later stages of the disease, full nursing care will be needed. Secondary illnesses, such as pneumonia, are often the actual cause of death.

What treatment and help is available?

Currently there is no cure for the illness, but there are many ways to manage symptoms effectively. Medication can be used to treat symptoms such as

involuntary movements, depression and mood swings. Speech therapy can significantly improve speech and swallowing problems. A high calorie diet can prevent weight loss and improve symptoms such as involuntary movements and behavioural problems. Social services in your local area can assist with practical issues such as appropriate adaptations to your home, where necessary, and they can also help with care at home or respite care. They can also assist with the provision of equipment, if necessary. A referral can be made through your local GP.

For more information about HD:

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)

Tel: 01224 552120 Fax: 01224 559390

(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)

Tel: 01382 632035 Fax: 01382 645731

(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)

Tel: 0131 651 1012 Fax: 0131 651 1013

(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)

Tel: 0141 201 0808 Fax: 0141 201 0361

(Glasgow, Argyll & Bute, Argyshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

Further information and support is available from:

Scottish Huntington's Association (Scottish HA)

Thistle House, 61 Main Road. Elderslie, PA5 9BA

Tel: 01505 322245

email: sha-admin@hdscotland.org.uk

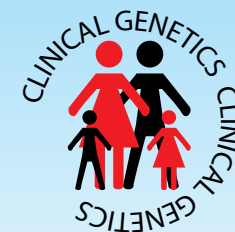
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What is Huntington Disease?



Information for families affected by Huntington Disease

What is Huntington Disease (HD)?

Huntington disease (HD) is a condition that affects the central nervous system. It is caused by a faulty gene on chromosome 4.

In some way - which is not yet understood - the protein product of the faulty HD gene is toxic to nerve cells in specific areas of the brain that control muscular movements, memory and mood.

DNA, Genes and Chromosomes.

DNA is like a coiled chain necklace where each link is made from one of four letters called nucleotides; adenine (A), cytosine (C), guanine (G), and thymine (T).

The order of the letters (e.g:CATGAGCTA) spells out to the cell the code for each of the 30,000 different genes that we have. Every gene has a unique code which is read as a triplet of letters (e.g: CAT GAG CTA).

A gene is a segment of DNA that has a particular purpose. A gene codes for (i.e. tells the body how to make) a specific enzyme or other protein. Genes determine personal characteristics such as eye colour and hair colour.

Our genes lie on tiny structures called "**chromosomes**", rather like beads (the genes) threaded onto a string (the chromosomes). Each chromosome contains thousands of genes.

- Most of our body cells have 46 chromosomes, arranged in 23 pairs.
- We inherit one of each gene pair from our mother and the other of each pair from our father.

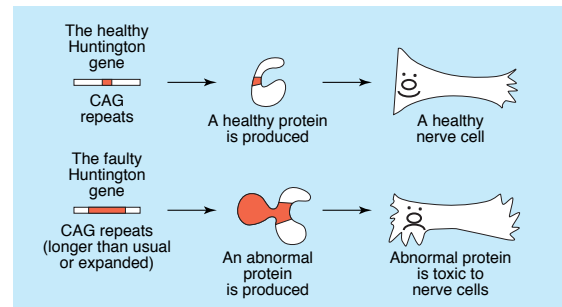
The HD gene

The HD gene, which codes for a protein called Huntingtin, was discovered in 1993.

Everyone has two copies of the **Huntingtin gene** and normally both genes have a series of CAG triplets, repeating 10-35 times, one after the other.

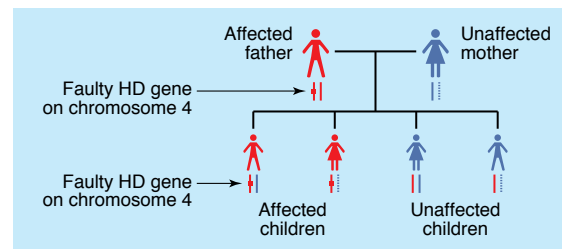
In someone who has HD the series of CAG repeats in one of their genes is expanded so that there are **more than 35 repeats** (usually between 37 to 100). Everyone who has HD will have one normal copy of the Huntington gene, inherited from their healthy parent, and one expanded copy with 37 or more CAG repeats, inherited from their affected parent. HD can affect both males and females. Males and females have the same chance of having affected children.

This increase in CAG repeats changes the shape and function of the Huntington protein and makes it toxic to certain nerve cells in the brain. It is not known how this occurs but a great deal of research is underway to find a treatment for HD.



Inheritance of the faulty HD gene

A parent affected with HD passes either the faulty HD gene, or the other working gene, to their offspring. There is a 50% (1 in 2) chance at each pregnancy that a child of an affected parent will receive the faulty HD gene. The age of onset, degree and type of clinical symptoms, as well as rate of progression varies with HD.



Genetic Testing

A genetic test is available for HD. This test will usually be able to show whether someone has inherited the faulty gene, but it will not indicate the age at which they will develop the disease, exactly how they will be affected, or how rapidly the symptoms will progress.

A variety of genetic testing options may be discussed with a Clinical Geneticist, Genetic Counsellor or Neurologist. These options might include direct testing for the faulty HD gene in an affected individual and subsequent testing of relatives. Genetic testing can be carried out by obtaining a blood sample. Most genetic testing and analysis should be done, ideally, before a family member begins a pregnancy. The decision by an individual to undergo genetic testing is an intensely personal decision and can be made only by that person.

How does one know if they have HD?

If a person has symptoms of clumsiness, poor balance or forgetfulness, a Neurologist or Geneticist, may be able to confirm a clinical diagnosis of HD by a simple examination. Unfortunately mild clumsiness and forgetfulness are common symptoms of stress and may be confused as signs of HD. A clinical diagnosis is usually not possible in most people who have no symptoms. If symptoms begin, however, and a diagnosis is made, each child of an affected parent has a 50% chance of inheriting the faulty HD gene.

Symptoms of HD

The symptoms of HD usually develop when people are between 30-50 years old, although they can start earlier or much later. The symptoms of HD can start at a different age and differ from person to person, even in the same family. Sometimes, the symptoms are present for a long time before a diagnosis of HD is made. This is especially true when people are not aware that HD is in their family.