NHS
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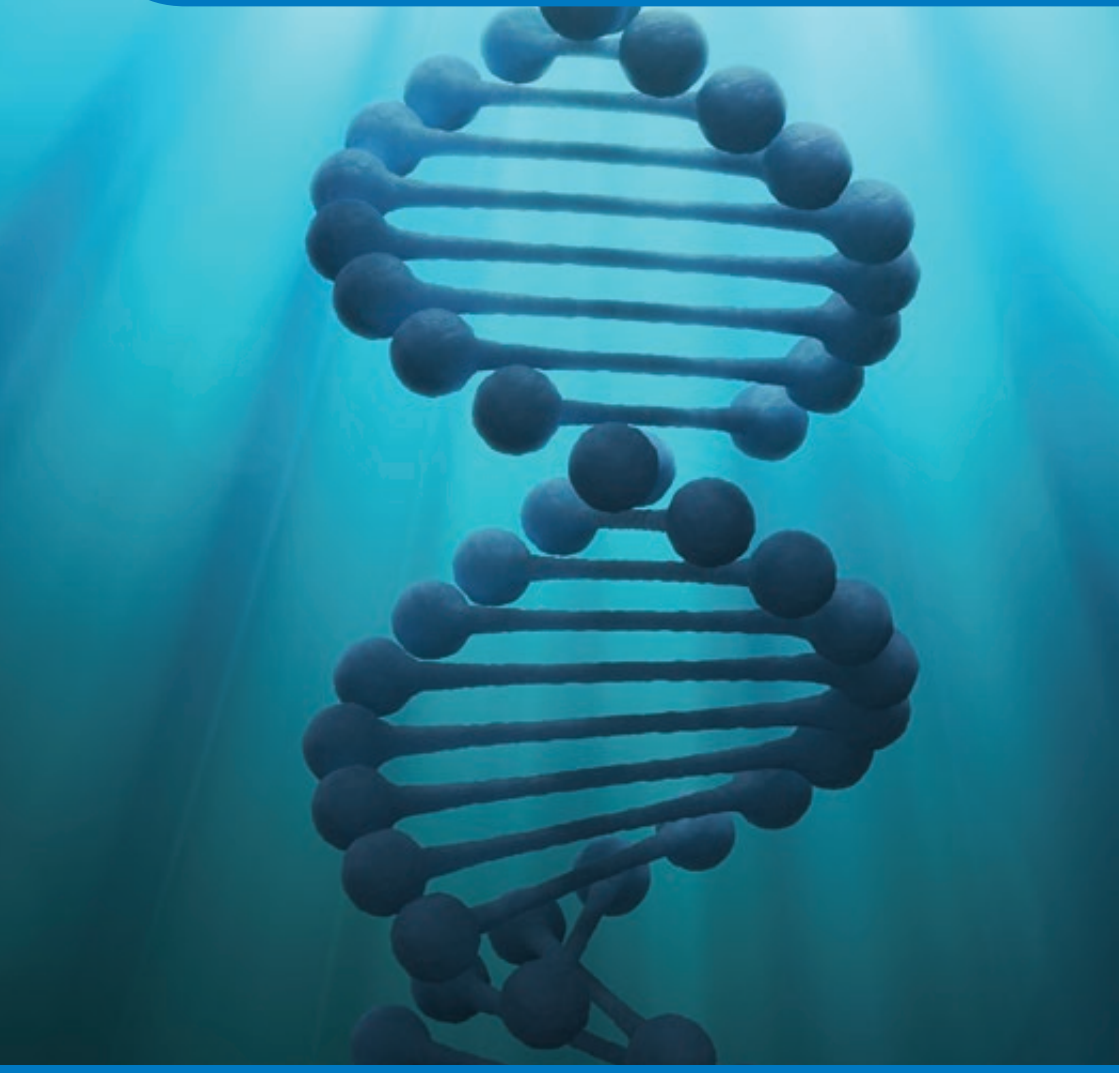
Huntington disease (HD)

Information for patients



We have written this booklet for people who have a family history of Huntington disease (HD) and would like to understand more about the condition. We hope this booklet will help to answer some of the questions you may have.

What is Huntington disease (HD)?

HD is a condition that affects the central nervous system and stops parts of the brain that control movements, memory and mood from working properly over time. It's passed on (inherited) from a person's parents. It is caused by a gene expansion on chromosome four.

Genes and chromosomes

Our genes can be thought of as a set of instructions for building and maintaining our bodies. There are around 25,000 different genes altogether and we have two copies of each one. 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 (for example, CATGAGCTA) spells out the code for each of the 25,000 different genes that we have. Every gene has a unique code which is read as a triplet of letters (such as CAT GAG CTA).

Our genes lie on tiny structures called chromosomes, rather like beads threaded onto a string. Each chromosome contains thousands of genes. Most people have 23 pairs of chromosomes (46 in total). One of each pair comes from your mother and the other from your father. Chromosomes are numbered one (the largest pair) to 22 (the smallest pair); the 23rd pair is known as the sex chromosomes, because they determine whether a person's sex is male or female. We only pass on one of each pair to our children.

The HD gene

The HD gene, which codes for a protein called Huntingtin, was discovered in 1993. Everyone has two copies of the HD gene and normally both genes have a series of CAG triplets, repeating between ten and 35 times, one after the other.

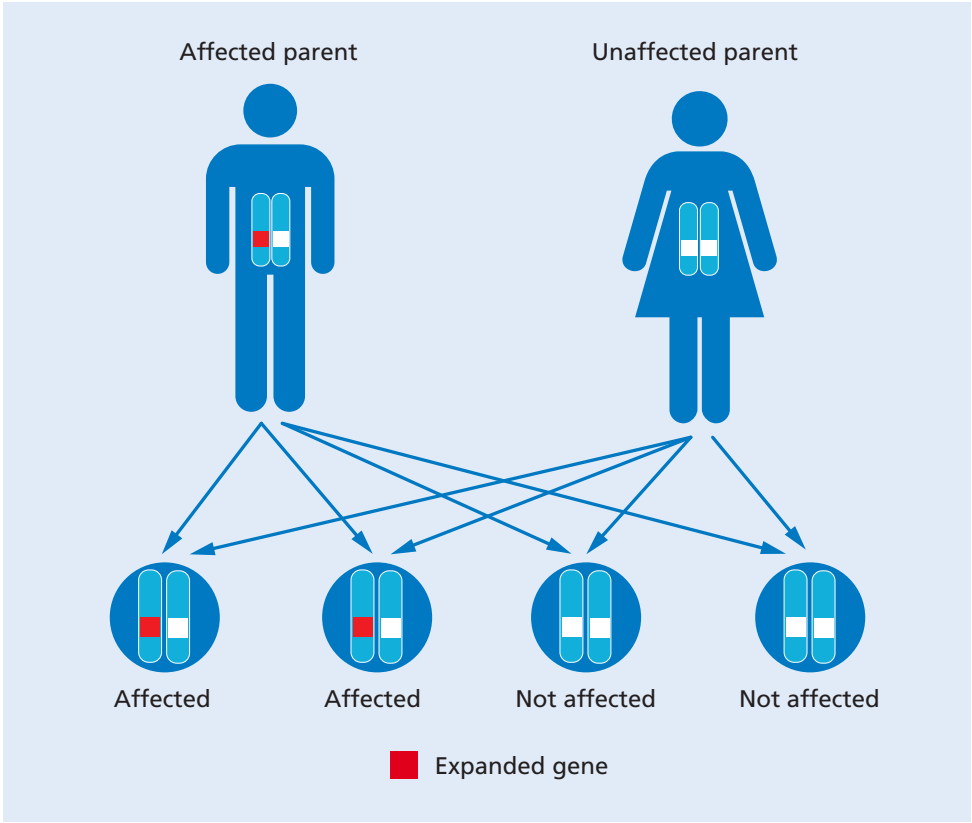
In someone who has HD, the series of CAG repeats in one of their copies of the gene is expanded so that there are more than 35 repeats (usually between 36 and 50). Everyone who has HD will have one normal copy of the HD gene, inherited from their unaffected parent, and one expanded copy with 36 or more CAG repeats, usually inherited from an affected parent.

This increase in CAG repeats changes the shape and function of the Huntingtin protein and makes it toxic to certain nerve cells in the brain. A great deal of research is underway to understand this and to try to find a treatment for HD.

Inheritance of the faulty HD gene

HD can affect both males and females. Males and females have the same chance of having affected children.

A parent affected with HD passes either the expanded HD gene, or the other working gene, to their children. There is a one in two (50%) chance in each pregnancy that a child of an affected parent will receive the expanded HD gene. The age of onset, degree and type of clinical symptoms, and progress of the disease varies in HD.



Testing

A genetic test is available for HD. This test will usually be able to show if someone has inherited the expanded 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. The decision to undergo genetic testing is personal and can only be made by that person.

Diagnosis

Symptoms of clumsiness, poor balance, forgetfulness or mood changes may lead a neurologist or clinical geneticist, after simple clinical examination, to suspect the onset of HD. Unfortunately, clumsiness, forgetfulness and mood changes are common symptoms of stress and may be mistaken for signs of HD. It may be difficult to be certain in the early stages whether someone is developing HD.

If the gene test is normal, a person cannot be affected by HD, nor can they pass an expanded HD gene onto their children.

Symptoms

The symptoms of HD usually develop between 30 and 50 years of age, although they can start earlier or later. Symptoms of HD can start at different ages and can 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. The early symptoms of HD include:

- lack of concentration
- short-term memory lapses
- depression
- changes of mood, sometimes including irrational or antisocial behaviour
- slight, uncontrollable muscle movements
- stumbling or clumsiness

Living with HD

As the disease progresses, symptoms may include:

- involuntary movements
- difficulty speaking and swallowing
- weight loss

Emotional changes are also common. These may result in:

- depression
- lack of insight (when a person is unable to recognise changes in their behaviour and emotions)
- sleep disturbance
- mood swings

Sometimes the psychological problems, rather than the physical deterioration, cause more difficulties for both the person with HD and the people who care for them. Loss of drive, initiative and organisational skills, may result in the person appearing lazy. There may be difficulty in concentrating on more than one activity at a time.

Some changes are definitely part of the disease process, although they may be made worse by other factors. For example, weight loss and depression are part of the disease but may be complicated by one another and by the increasing physical difficulties the person with HD experiences.

In the later stages of the disease, full-time nursing care will be needed.

Treatment

Currently there is no cure for HD, but there are many ways to manage the symptoms:

- Medication is available for symptoms such as involuntary movements, depression and mood swings.
- Speech therapists can help with speech and swallowing problems.
- A high calorie diet can help prevent weight loss and improve symptoms such as depression and behavioural problems.
- Occupational therapists can assist with practical issues such as appropriate adaptations to the home and provision of equipment.
- Social services can help with care at home or respite care (a short-term break for caregivers).
- Financial benefits are available to support affected individuals and their carers in coping with the extra expense of illness.

Contact us

If you have any questions or need advice about any aspect of HD, please contact us:

- **Wessex Clinical Genetics Service**
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  Telephone: **023 8120 6170**
  Website: **www.uhs.nhs.uk/genetics**

Further information

- **Huntington's Disease Association (HDA)**

  Telephone: **0151 331 5444**
  Email: **info@hda.org.uk**
  Website: **www.hda.org.uk**

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