

Huntington's Disease - *Factsheet*

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Information resource for carers, professionals and you

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Huntington's Disease

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Dementia is not an illness in itself, but a term for a collection of conditions and diseases which cause disorders in the brain. One of the rarer conditions that can cause dementia in younger people is Huntington's Disease (also known as Huntington's Chorea).

What is Huntington's Disease

Huntington's disease is an hereditary neurological disorder of the central nervous system. It was first described by George Huntington, an American doctor, in 1872. He called the disease "chorea" from the Greek word meaning dance, because of the rapid, jerking uncontrollable movements which occur. The disease therefore came to be known by its former name of Huntington's Chorea.

Huntington's disease is a genetic disorder, caused by a single defective gene on chromosome 4. It is estimated that between 6,500 and 8,000 people in the UK have the disease. Each person who has a parent with the disease has a 50% chance of developing the disease themselves. As the symptoms of Huntington's disease usually develop when people are between 30-50 years of age, and if they are unaware that it is an inherited condition, they may well have already passed it on to their children. This can happen because the disease was not diagnosed in previous generations or was wrongly diagnosed, perhaps as mental illness.

How Huntington's disease affects the brain

The defective gene (which was only discovered in 1993) causes damage to the nerve cells in certain areas of the brain and spinal cord. Abnormal brain cells are found in those deep areas of the brain which control movement. A result of the abnormal gene, a protein named huntingtin, is thought to prevent brain cells from protecting themselves against oxidants which are naturally produced in the body. Research is also going on to investigate whether some neurotransmitters in the brain are dying because they are unable to use energy efficiently. Some experimental treatments have aimed to slow the disease by providing added protection against oxidants.

The symptoms of Huntington's disease

Symptoms usually develop gradually, starting between about thirty and fifty years of age, although in about 5% of people symptoms can appear before the age of twenty. Symptoms may also not appear until well past fifty.

Early symptoms are slight, and they can be present for a long time before a definitive diagnosis is made. Sometimes, changes in personality or mood might be the first sign of the disease, and this could happen as much as ten years before other symptoms appear. When other symptoms start to be seen, there might be slight, uncontrollable muscle movements, occasional memory loss or confusion, clumsiness, unexplained falls, and lack of concentration.

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The abnormal movements which are characteristic of the disease are usually, at the outset, rapid, jerking movements which can involve all four limbs, the trunk, the face and the head. As the disease develops, strange postures and a general slowing of movement may occur. The person may develop a lurching gait, and be generally unstable and prone to falls. Speech may also be affected, and it may become difficult to understand what the person is saying. The loss of muscle control and co-ordination, the inability to close the lips and irregular movements of the diaphragm can make eating very hard, and even dangerous, as a difficulty with swallowing makes choking a considerable risk. People commonly lose weight, and this is thought to be due partly to burning more calories with the continuous movement and partly to the difficulties with eating. Some people experience problems moving their eyes quickly and also with looking sideways – they may need to move their whole head.

Emotional changes are very common, and people with Huntington's disease are often described as "stubborn". They may be frustrated, depressed – and there is a higher incidence of suicide than in the general population. These changes are not only due to the obvious stress of having the disease, but are also a direct result of the damage to the brain. Later in the disease people may develop symptoms similar to those of Parkinson's disease, ie slowness of movement and rigidity. Other symptoms may include a loss of drive and initiative, and an inability to plan, organise, or do more than one task at once. In the later stages of the disease, memory loss is likely to become worse, and the person may find it difficult to understand what people are saying, become less mobile, and more self-centred.

Huntington's disease often progresses very slowly, and the affected person may live for fifteen to twenty years after diagnosis. Eventually, full-time nursing care will be needed. Secondary illnesses, such as pneumonia, are often the cause of death.

What treatment can be given?

There is currently no cure for Huntington's disease, but there is much that can be done to ease the symptoms and help the person and their family to lead a normal life for as long as possible. There are a variety of drugs which can be used, for example, to reduce the involuntary movements, to alleviate depression and to stabilise mood. It is important to try to ensure that the person has a high calorie diet. This can help to prevent weight loss and can also improve other symptoms. Speech therapy can help to improve speech and eating and swallowing problems, dieticians can advise on adequate intake and counselling can help both the person and their family.

Although there is no cure at present, scientists are currently investigating drug treatments which might slow the progression of the disease. There are plans to test some of these in clinical trials. However, at the moment, the research is speculative and much more work is needed. Another area of research is the use of transplants of stem cells, but this is in the very early stages of development.

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What to do

Many people with Huntington's will already be aware that they may have inherited the disease. This means that if they decided not to undergo testing they may nevertheless spend years worrying, and looking out for likely symptoms. They could be worrying unnecessarily, as the early symptoms, such as stumbling, dropping something, forgetting someone's name or being bad-tempered could happen occasionally to anyone.

However, if it becomes apparent that something is amiss, at some point it will become necessary to seek a diagnosis. If the person has a family history of Huntington's disease and starts to show symptoms which do not seem to be attributable to any other disease, the diagnosis of Huntington's will usually be clear. Otherwise, there are tests available which might reveal some of the abnormalities in memory and thought processing. An MRI scan may identify typical shrinkage in the brain, and other scans may confirm a diagnosis, as may genetic testing for the abnormal gene.

Genetic testing

Huntington's disease is hereditary and tests are available which can establish whether the faulty gene is present. If someone knows they have the faulty gene, their unborn children can be tested to see if they have inherited it. They may also have access to IVF, using mutation-free embryos.

A person who knows their parent has tested positive is in a very difficult position. Because there is no cure, and furthermore, no way of predicting when the disease might manifest itself, or how quickly it might progress, it is questionable whether it is a good idea to take the test. A positive result will be very distressing, and may have serious psychological effects. In addition, it may be that the parent has decided not to be tested, so if their child is found to carry the gene it will have severe implications for them too.

In the UK you cannot have this test until you are 18 years old, and in any event you should certainly sort out life insurance before considering it, as it may be very difficult to get this if you subsequently test positive. Testing is available at Regional Genetics Clinics and you will be required to engage in a counselling procedure over at least three sessions. You should be aware that in a few cases testing will not produce a definitive result, and the whole process will take weeks or months, and will, obviously, be very stressful.

Where to get help

Contact your GP for support, advice and information. The GP will rule out reversible or temporary causes for symptoms, carry out first line tests, refer to a specialist for specific tests and assume ongoing responsibility for the person's general health. The GP can be seen as a "gatekeeper" who can provide access to a range of specialist services. Home support, day services and 'talking therapies' may be provided.

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Many other people may also be able to help, such as social workers, community psychiatric nurses, district nurses, speech therapists, occupational therapists, counsellors, advocates and carers who may come into the home to assist with personal care.

It may be that the person's illness reaches a stage where their family can no longer cope with their care. If moving into a care home is seen to be in the person's best interests, independent reports and quality ratings on all homes in your local area can be found on the website of the Care Quality Commission which regulates them. www.cqc.org.uk (use its webform)

Or telephone 8.30 am to 5.30 pm Monday to Friday on **03000 616161**

You can also refer to our factsheet: "Moving to a Care Home and Funding Your Care".

Guideposts Trust provides specialist information and care services for people with dementia and their carers. www.dementiaweb.org.uk

Contact the Helpline number: **0845 1204048** available Monday to Friday office hours, answer service at other times or by email at info@dementiaweb.org.uk

The Huntington's Disease Association has a very comprehensive website, with free, downloadable factsheets and booklets on every conceivable aspect of Huntington's. There are branches and support groups around the country and a network of regional care advisors.

The website can be found at www.hda.org.uk or make contact at:

Neurosupport Centre, Norton Street, Liverpool, L3 8LR Telephone: **0151 298 3298**

Fax: **0151 298 9440**

Email: info@hda.org.uk

Carers UK provides advice and information to carers and the professions who support carers.

Carers UK Adviceline: **0808 808 7777** (Wednesday and Thursday 10am to 12pm and 2pm to 4pm)

Email: adviceline@carersuk.org Website: www.carersuk.org

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Dementia Information Service for Carers

Helpline Number **0845 1204048**

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Email: info@dementiaweb.org.uk

Web: www.dementiaweb.org.uk

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