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Huntington's Disease

Huntington's disease is an inherited (genetic) condition that affects the brain and nervous system. It is a slowly progressive condition that interferes with the movements of your body, can affect your awareness, thinking and judgement and can lead to a change in your behaviour.

The symptoms occur because of damage and death of some of the brain cells (neurons) in particular parts of your brain. Genetic testing helps to diagnose Huntington's disease.

At present there is no cure for Huntington's disease. Treatment is aimed at trying to control symptoms as much as possible when they develop.

What is Huntington's disease?

Huntington's disease (HD) is named after George Huntington who first described it in 1872. It is an inherited (genetic) condition that affects the brain and nervous system. It can interfere with movements of your body, can affect your reasoning, awareness, thinking and judgement (cognition) and can lead to a change in your behaviour. 'Genetic' means that the condition is passed on through families by special codes called genes. Each cell of your body contains chromosomes which are made up of many genes.

What causes Huntington's disease?

HD is caused by a defective gene that you inherit from your parents. This faulty gene is carried on chromosome 4. The gene is responsible for making a protein called 'huntingtin'. The genetic defect means that certain proteins needed to make brain chemicals cannot be made in your brain as normal. It is thought that this leads to damage and death of some of the neurons (brain cells) in particular parts of your brain called the basal ganglia and the cortex. It is this damage that leads to the symptoms of HD. There is also a build-up of a chemical called dopamine in the brain which contributes to the problems with moving.

HD is an autosomal dominant condition. This means that you can inherit HD from just one of your parents. If one of your parents has a faulty copy of the gene, there is a 50:50 chance that each child they have will inherit the faulty gene and develop HD. See the end of this leaflet for a diagram to help you to understand this.

However, occasionally, someone with HD may not have a history of HD in their family. This may be because of what is called a 'new mutation' (a 'de novo mutation'). A new mutation is a mutation (or a fault) in a gene that is present for the first time in one family member. It can happen because of:

- A fault in the genetic material in either the egg or sperm of one of the affected person's parents; **or**
- A fault in the genetic material of the egg fertilised when the egg and sperm meet (the embryo).

It is not clear what causes this fault (mutation) to suddenly occur for the first time.

Also, if an individual in your family who is carrying the faulty HD gene dies before their symptoms develop (and therefore before HD is diagnosed), relatives will not be aware of this family history. This can be another reason for a lack of history of HD in your family.

How common is Huntington's disease and who develops it?

HD affects between 5-10 people per 100,000 in the UK. Worldwide, it seems to be more common amongst white populations than amongst Asian or African people. HD affects both men and women equally. It is most common to start to develop the symptoms of HD between the ages of 30-50 years. So, even though you may have inherited the disease, it can take some time to develop the symptoms.

Between 5-10 in 100 people with HD develop the symptoms before they are 20. This is known as juvenile-onset HD. It can cause more severe symptoms. Most people who have this type of HD inherit the condition from their father.

What are the symptoms of Huntington's disease?

The symptoms of HD can vary from person to person. Not everyone with HD has the same symptoms or develops every symptom. The symptoms tend to progress gradually and worsen over time.

If HD does not run in your family, it can sometimes take a while for it to be diagnosed. This is because some of the early symptoms can often be overlooked or put down to other problems - for example, subtle changes in your personality, or mood swings.

The symptoms of HD can be grouped into three main areas:

- Problems with movement.
- Problems with cognition.
- Mood and behavioural problems.

Problems with movement

The most common movement problem in HD is known as chorea. In fact, until recently, HD was known as Huntington's chorea. Chorea means a jerky, involuntary movement. In particular, it tends to affect your head, face, arms or legs. Each movement happens suddenly. In the beginning some people may just think that you are fidgety; however, your symptoms tend to gradually worsen. In severe chorea, you can develop uncontrollable flinging (or flailing) of your arms or legs (called ballism). This can interfere with your ability to move around. You may be more likely to fall and have difficulty, for example, feeding or dressing yourself. You may drop things easily.

Over time, as HD progresses, you can develop a movement problem known as dystonia. This tends to gradually replace chorea. Dystonia means you have spasm in your muscles, usually the muscles of your shoulders, neck, arms and legs. This can lead to twisting movements, repetitive movements, or abnormal postures. You may also notice that over time your limbs can become quite stiff or rigid and your movements tend to slow down. This slowing of movements is called bradykinesia.

Swallowing problems are common if you have HD. This is because HD can affect the muscles that control your swallowing and the muscles around your mouth. Choking can be a problem and also difficulty eating means that you can become underweight.

You may notice problems with your speech and your speech can become slurred. Your eye movements may also be affected, causing problems looking from side to side or up and down.

Problems with cognition

Your cognition (your perception, reasoning, awareness, thinking and judgement) is closely linked to your behaviour. So, changes in your cognition due to the loss of neurons in your brain can also affect your behaviour (see below).

The first symptoms that you may notice are problems with your short-term memory. Lack of concentration, short-term memory lapses and problems with orientation are common. Learning new skills can become difficult.

The slow decline in cognition in HD can be similar to a dementia-type problem.

Mood and behavioural problems

Changes in your behaviour can be one of the first signs of HD. They may come on before your movement is affected. However, in some people, they start after the onset of movement problems.

You may notice that you become irritable, easily agitated, and start to lose interest in things that you previously enjoyed. People may comment that you are becoming untidy and you may start to lose interest in your self-care - for example, washing less frequently, not taking care of your appearance, etc.

Your judgement about things can become affected. You may become aggressive towards people and you can sometimes lose your inhibitions, leading to embarrassing behaviour in social situations. The personality changes related to HD can be very difficult for you or your family and friends to deal with. Behavioural changes and antisocial behaviour can put a strain on your relationships. You may find it difficult to accept that your behaviour may be a problem. It is all part of the way that HD affects your brain.

Depressed mood is common in people with HD and there is an increased risk of suicide. Other mental health problems, including obsessive-compulsive disorder and problems similar to mania and schizophrenia, are also more common in people with HD. See the separate leaflets called Obsessive-compulsive Disorder, Bipolar Disorder and Schizophrenia for more details.

Do I need any investigations?

If you already have symptoms

Genetic testing is available to confirm HD. However, if you already have a family history of HD and you have the typical symptoms and signs of the condition, genetic testing may not be necessary.

A CT scan or MRI scan of your brain may show some typical signs of HD. However, these scans are not usually helpful when trying to diagnose HD in its early stages as changes may not be present. Scanning is mostly used in the later stages of the disease.

If Huntington's disease runs in your family

If there is HD in your family, it can cause a lot of worry and anxiety, as you may be looking out for possible symptoms. If you are at risk of developing HD because of a family history but you have not yet developed any symptoms, genetic testing is also possible. This is known as pre-symptomatic testing. If you have inherited the defective gene, you will definitely develop symptoms at some point in your life. However, it is not possible to predict when your symptoms will appear.

Whether or not to have genetic testing is a choice for each person as an individual. Some people do not wish to be tested. They would prefer to wait to see if they develop any symptoms. It is recommended that you undergo counselling if you are considering genetic testing. Your GP will be able to refer you to a specialist who is a genetic counsellor.

What is the treatment for Huntington's disease?

At present there is no cure for HD. Also, there is no treatment that has been found to delay the onset of symptoms or to delay the progression of symptoms. So, treatment is aimed at trying to control symptoms as much as possible when they do develop.

Treatments with medicines

There are a number of different medicines that can be used to help treat chorea. A group of medicines known as benzodiazepines is often used first. Examples of these medicines include clonazepam and diazepam. Other medicines, including tetrabenazine, may also be used. However, all these medicines have possible side-effects. For example, they can lead to slowing down of your movements (bradykinesia), stiffness or rigidity, depression or sedation. Whether and when to start treatment will depend on striking a balance between benefits and side-effects.

If you have problems with bradykinesia and stiff or rigid limbs, a group of medicines called dopamine agonists may be helpful. Another medicine called levodopa is an alternative. These medicines are commonly used to treat Parkinson's disease where there is similar slowing of movements and rigidity.

If you develop depression, antidepressant medicines can be helpful. Medicines are also available to treat some other mental health problems that may be associated with HD.

Other treatments

Most people with HD have a team of healthcare specialists who work with them. Your GP often co-ordinates your care. Other team members may include a specialist in brain, nerve and muscle problems (a neurologist), a psychiatrist and a genetic counsellor.

You may also be referred to a physiotherapist for help with exercises for your balance and exercises to help you move around more easily. An occupational therapist may be able to help you with any adaptations that you need to make your day-to-day life easier. For example, they can help with adaptations to your home such as wheelchair access, rails and changes to your bedroom and bathroom.

A speech and language therapist may be able to help with speech and/or swallowing difficulties. They may be able to teach you different ways of communicating. You may be referred to a dietician if you lose a lot of weight (due to swallowing difficulties and also your movement problems). They can advise about foods that may be easier for you to eat because they involve less chewing. Sometimes swallowing problems can mean that you need to consider having a nasogastric tube fitted. This is a tube that passes through your nose to your stomach so that food can be delivered to your stomach without you having to swallow.

Possible future treatments

Various new treatments for HD are being studied. They include gene therapy treatments and various treatments with medicines. For example, trials looking at the effect of a new medicine called pridopidine are underway. Trials are also looking at medicines to prevent people who have the faulty HD gene from developing the disease. However, these treatments are still very much at an experimental level. More research is needed before we will know whether such treatments are helpful for HD.

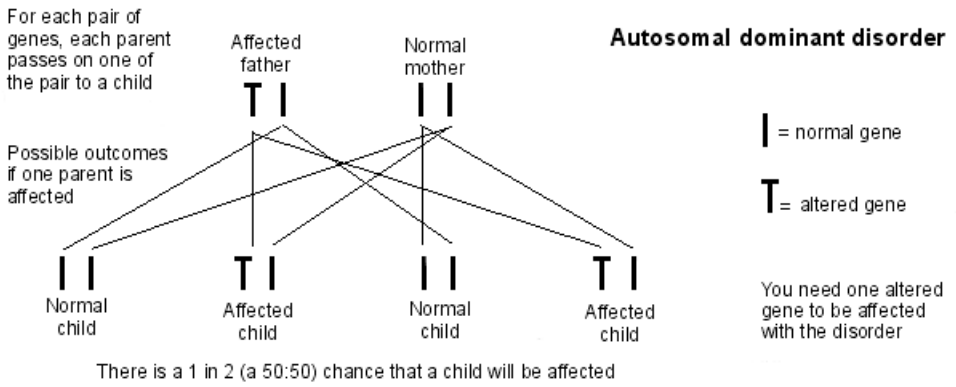
What is the outlook (prognosis) with Huntington's disease?

HD is a condition that slowly progresses so that you gradually develop more, and worsening, symptoms. In the later stages of HD, you will become totally dependent on other people and require full nursing care. HD leads to considerable disability and, at present, will eventually lead to death.

At present, most people live from between 10 to 25 years after they first develop the symptoms of HD. Someone with HD usually dies from an infection such as pneumonia but suicide is very common. Remember that new treatments are under investigation. Your specialist and the Huntington's Disease Association will be able to discuss any treatment developments or trial treatments with you.

Can I pass Huntington's disease on to my children?

People with HD often have children before developing symptoms of the disease. If you carry the defective HD gene, for each child that you have, there is a 50:50 chance that they will also have HD. If you or your partner have HD, prenatal testing is available. This can show whether your baby has the defective gene and therefore whether they will develop HD. However, testing is not always 100% accurate.



Preimplantation genetic diagnosis (PGD) is also available if one parent carries the defective gene. This basically involves the couple undergoing IVF-type treatment so that embryos can be tested for HD before they are implanted in the woman's womb (uterus). Only embryos without the defective HD gene are implanted.

Genetic counselling and specialist advice are recommended if you or your partner have HD, or if there is HD in either of your families, and you are considering pregnancy.

Further reading & references

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