

Is a genetic test available for HD?

A blood test can show whether someone has inherited the faulty gene but will not be able to predict the age at which they will become affected, exactly how they will be affected, or how quickly the symptoms will progress. There are many issues to consider before undergoing a genetic test. These can be discussed with a Genetic Counsellor. The decision to undergo genetic testing is a very personal decision which can only be made by the individual themselves. Genetic testing is not usually offered to people under the age of 18.

Tests during pregnancy can be offered to gene carriers who want to find out if the pregnancy is affected. These tests are complex and a Genetic Counsellor or Clinical Geneticist can provide more information. These tests are ideally discussed before beginning a pregnancy but if this is not possible, they should be discussed as early in the pregnancy as possible.

For further information and support about HD you could contact the Huntington's Disease Association (HDA) at:

Huntington's Disease Association
Suite 24 , Liverpool Science Park
Innovation Centre 1
131 Mount Pleasant
Liverpool
L3 5TF

Telephone: 0151 331 5444
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Website: www.hda.org.uk

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Birmingham Women's 
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Huntington Disease (HD)

An information leaflet for patients and families

If you need more advice about any aspect of Huntington Disease please contact:

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Introduction

Huntington Disease (HD) is a disorder of the central nervous system. It used to be known as Huntington's Chorea. HD runs in families because it is caused by a faulty gene which can be inherited by males and females. It affects muscular movements, memory and mood. The illness begins very gradually and the condition slowly progresses.

Early symptoms of HD

The age at which symptoms develop can vary from one person to another. The onset of the condition is usually between the ages of 35 and 55 but may be earlier or later. Even within the same family the age of onset can vary from one person to another. Early symptoms include stumbling, clumsiness and slight, uncontrollable muscular movements. Lack of concentration, short term memory lapses, depression, mood changes, irritability, aggression and antisocial behavior can all be early symptoms of HD. The onset of symptoms can be subtle and they may be present for some time before a diagnosis of HD is made.

How HD progresses

Later on in the illness people may experience many different symptoms including involuntary movements, weight loss and difficulty with speech and swallowing. The emotional changes can lead to stubbornness, frustration, mood swings and depression. Some people experience changes in the way that they think which are often referred to as 'cognitive' changes. These cognitive changes

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

Sometimes these psychological problems cause more difficulties, both for the person with HD and their carers, than the physical deterioration. Some changes are definitely part of the disease process but may be made worse by other factors. For instance, 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 illness full nursing care will be needed. Secondary illnesses, such as pneumonia, are often the actual cause of death.

What treatment and help is available?

Although there is currently no cure for HD there are treatments which can help to manage the symptoms. 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 may also improve symptoms such as behavioural problems. There are also adaptations which can be made within the home environment to make day-to-day living easier.

What causes HD?

HD is caused by a faulty gene. Genes are the units of information which tell the cells of our bodies how to work. The HD gene produces a protein which is not

usually harmful. However, when there is a fault in the gene it produces an altered protein which can damage certain nerve cells in the brain. We know that there is a part of the genetic code within the gene where three letters of the genetic code repeat themselves. When the gene is faulty, the number of times that these letters repeat themselves is greater than within the general population. This is called an 'expansion' in the repeat size.

How is HD inherited?

We all have two copies of the gene because we inherit one copy from each of our parents. The HD gene is a dominant gene. This means that if someone inherits one faulty copy of the gene they will develop symptoms of HD at some point in their lifetime. If someone has a faulty copy of the gene, each of their children will have a 1 in 2 (50%) chance of inheriting the faulty copy of the gene. This is shown in the diagram below.

