

Huntington Disease (HD)

What Is Huntington Disease?

Huntington Disease (HD) is a genetically inherited disorder which involves gradual loss of nerve cells in the brain. It is sometimes referred to as Huntington's chorea due to abnormal movements of the arms and legs (chorea). HD is named after Dr George Huntington who recognised that it was an inherited condition in 1872.

Huntington Disease is often diagnosed through a careful family history. Brain scans and genetic testing are also used to confirm the diagnosis. The Division of Human Genetics at the University of Cape Town currently tests for HD.

What is the genetic fault that causes HD?

HD is caused by a genetic mistake that involves the repetition of a particular DNA region over and over again. The scientists refer to this as a 'triplet repeat' expansion.

What are the symptoms of HD?

The age at which symptoms first appear varies but it has been found that the longer the 'triplet repeat', the earlier in life the symptoms start. Generally the first symptoms become apparent between the ages 35 and 50, appear slowly and are often not noticed by the person. In the early stage of Huntington disease, most people can still enjoy many of their normal activities, including going to work. Involuntary movements such as twitching of the fingers and toes are infrequent, speech is unaffected and dementia, if present, is mild. Later, patients need more assistance with daily living. The symptoms are usually described as "clumsiness", "jerkiness", "tremor" or "balance trouble". Personality changes including irritation, poor insight, depression, withdrawal, euphoria and difficulty with organisation may also be noticed. As the disease progresses, involuntary movements become more obvious. Difficulties with speaking and swallowing will develop and the person will walk in a wide-based, unsteady way. Falling and weight loss are common. Eventually reasoning and judgement

become impaired and dementia is more obvious. In the late stage of the disease, patients need almost total care. They may be unable to walk or speak, and rigidity (stiffness) may become more common than involuntary movements.

The person with HD eventually succumbs to pneumonia or complications of falls or choking. Life expectancy is usually about 15-20 years after the disease starts.

How Is HD Inherited?

Huntington disease is inherited as a dominant trait, which means that any parent who carries the fault in the gene will have a 50% chance of having an affected child. (see Autosomal Dominant sheet). However, if a child does not inherit the gene it cannot pass it on to its own children. The chain of inheritance is then broken.

It has been shown that the 'triplet repeat' tends to expand (become longer) from one generation to the next. This is referred to as 'anticipation' and means that symptoms may start earlier in life and be more severe when the altered gene is passed on to the next generation.

How common is HD?

Huntington disease affects about 1 in 10 000 people. It affects men and women equally and occurs in people of all ethnic origins.

Is there any treatment or cure for HD?

Not at present. There is also no treatment to slow the progression of the disease. Treatment is aimed at supporting the patient and his/her family. Prescription drugs can help control some of the symptoms, especially the involuntary movements and psychiatric conditions such as depression.





Division of Human
Genetics
University of Cape
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Testing

The NHLS provides genetic testing for people affected with HD (diagnostic testing) and for their family members if they are over 18 years old (predictive testing). This test accurately determines if a person has inherited the mutated (faulty) gene that causes HD. A positive result for the mutation means that the person will develop HD at some point in their lives. There is however no way of predicting when or how the condition will start or progress.

HD Predictive Testing Protocol

The predictive test for Huntington disease allows testing of family members that may be at risk, before any clinical signs present. A programme or protocol has been recommended by doctors and geneticists based on their experiences in dealing with individuals and families at risk for the disease. If, after careful consideration, you decide to take the test, you will be requested to come to the Groote Schuur Hospital Neurogenetic Clinic or the Department of Human Genetics on at least four occasions to see the doctors involved in running the programme.

The full protocol for predictive testing is available at the following website:

<http://www.humangenetics.uct.ac.za>

How soon will results be available?

Results will be available within approximately 4 weeks of the test. The results will be given to you personally via your general practitioner, neurologist or by the staff of the Division of Human Genetics at the University of Cape Town.

Genetic counselling

As this is a genetic condition, genetic counselling is strongly recommended. Genetic counselling provides information on the condition, its inheritance pattern, risks to other family members and the prognosis. The Division of Human Genetics at the University of Cape Town (UCT) can be contacted in this regard.

Further Information On HD Can Be Found At:

<http://www.humangenetics.uct.ac.za>

<http://geneclinics.org/profiles>

Contact Details:

A Neurogenetic Clinic is held at Groote Schuur on the first Thursday of every month.

Tel: (021) 404 6235 or 406 6304

Support Groups:

HASA support group in Cape Town

www.huntingtons.org.za

Abbotts College, Greenwood Rd, Claremont, Cape Town 7708

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