Anyone considering taking the test should have genetic counseling. This will ensure that he or she understands what the possible outcomes could be, and whether the decision to be tested is the right one at that time.

Are there any treatments?

In 2006, Canadian researchers provided bold new hope when they were able to successfully cure the disease in mice. This groundbreaking research is the first of its kind in the world and represents a major milestone on the path to the discovery of an effective treatment and a cure. Testing and further study of this new model of prevention are ongoing.

At the moment, however, there are notreatments that will slow down or stop the disease in humans. There are some drug treatments available that can reduce some of the symptoms of HD, such as depression, anxiety, and involuntary movements. These drugs can have side effects, so not everyone with Huntington's uses them. On a more promising note, there are several advanced drug trials underway. Scientists are extremely excited about the hope that these drugs may hold.

Researchers are also looking at surgical treatments, such as implanting fetal brain cells into the brains of Huntington's patients in the hope the cells will grow and take over the functions of the dead cells.

In addition, neurologists, psychologists, genetic counselors and social workers can play an important role in helping individuals or families deal with the disease. Physical therapists, occupational therapists and speech therapists can also help people with Huntington's cope better with some of the symptoms.

And because people with HD often lose a lot of weight, a nutritionist can be very helpful. It is important that all of these professionals work together to help manage the most effective treatment for each individual, since the disease often develops differently in different people.

The Dutch Huntington Association

The Dutch Huntington Association is a National network of volunteers and professionals working to find new treatments and ultimately a cure for Huntington disease, and to improve quality of life for people with HD and their families.

Your care is our care!

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What is Huntington Disease?

A brief description

What is HD?

Huntington disease (HD) is an inherited brain disorder. It was named after the doctor who first described it in 1872 – George Huntington – and it used to be called Huntington's chorea. HD causes cells in specific parts of the brain to die: The caudate, the putamen and, as the disease progresses, the cerebral cortex.

As the brain cells die, people with Huntington's become less able to control movements, recall events, make decisions and control emotions.
The disease leads to incapacitation and, eventually, death.

Who gets it?

Huntington disease is a genetic disorder. In the Netherlands there are about 1.700 people with HD and approximately 6.000 to 9.000 people are at risk of developing the disease. The HD gene is dominant, which means that each child of a parent with HD has a 50% chance of inheriting the disease and is said to be "at risk".

Males and females have the same risk of inheriting the disease. Huntington's occurs in all races. Primarily, HD affects adults. Symptoms usually appear between the ages of 30 and 45, but the disease can first appear in children as young as 5, or in adults in their 70s.

What causes it?

It's not clear how the abnormal HD gene causes the disease, Since the discovery of the gene in 1993, scientists have been working hard to discover the biochemical processes that causes the brain cells to die.

So far, we have learned that the HD gene produces a protein called "huntingtin". In people with HD, this protein gets cut into one short piece and one longer piece. The shorter pieces stick together to form a protein ball. Scientists are currently investigating whether it is the breakage of the protein, the formation of protein balls, or some other process that leads to cell death. They are also trying to understand why only certain brain cells die.

What are the symptoms?

There are three main types of symptoms in Huntington disease:
• Physical symptoms, including involuntary movements and diminished coordination
• Emotional symptoms, including depression, irritability and obsessiveness
• Cognitive symptoms, including loss of ability to recall information, loss of attention and difficulty with decision making.

There is a lot of variation in symptoms, and not every person will have all the symptoms to the same degree. Symptoms also vary witheach stage of the disease.

Early Stage
Early symptoms of the disease often include subtle cognitive changes. People with early Huntington's may find they have difficulty organizing routine matters or coping effectively with new situations. Difficulty recalling information may make them appear forgetful. Work activities may become more time-consuming and decision making and attention to details may be impaired.

Early emotional symptoms may be equally subtle and can include irritability or impulsiveness, or depression. Slight physical changes may also develop at this stage. There can be involuntary movementswhich may initially consist of "nervous" activity, fidgeting, a twitching of the hands or feet, or excessive restlessness. Individuals may also notice a little awkwardness, changes in handwriting, or difficulty with daily tasks such as driving. At this stage, people with Huntington's can function quite well at work and at home.

Intermediate Stages
As the disease progresses, the symptoms become worse. The initial physical symptoms will gradually develop into more obvious involuntary movements such as jerking and twitching of the head, neck and arms and legs. These movements may interfere with walking, speaking and swallowing.

People at this stage of Huntington's often look as if they're drunk: they stagger when they walk and their speech is slurred. They have in creasing difficulty working or managing a household, but can still deal with most activities of daily living.

Advanced Stages
The advanced stages of Huntington's typically involve fewer involuntary movements and more rigidity. People in these stages of HD can no longer manage the activities of daily living and usually require professional nursing. Difficulties with swallowing, communication and weight loss are common.

Death usually occurs 15 to 25 years after the onset of the disease. People don't die from Huntington's itself, but from complications such as choking, heart failure, infection or aspiration pneumonia.

Juvenile HD

Close to 10 percent of Huntington's cases are considered "juvenile" – that is, the symptoms occur in childhood or adolescence. The symptoms of juvenile HD are somewhat different from the adult disease. Children with HD move slowly and stiffly, they have increased difficulty learning and they can have convulsions or epileptic seizures. Some children have severe behavioural problems. Because these symptoms can be very different from those in adults, it can be difficult to diagnose.

How is HD diagnosed?

Huntington's is usually diagnosed using neurological and psychological tests, and with a review of family history. Sometimes doctors use brain scans to see whether the caudate and putamen are working properly, or they use the genetic test (see below) to confirm the diagnosis.

Genetic Testing

Since 1986, genetic testing for HD has been available; however, in 1993, a direct test for the disease was developed. This means people who are at risk for Huntington's or who believe they have the symptoms can take a blood test to tell them whether they have the gene that causes HD.

Many people at risk choose not the take the test. One reason may be that there is still no treatment to prevent HD from developing if the gene is present. But some people want to know whether they will eventually get the disease, so they can make arrangements as far as careers, insurance, family planning and other issues are concerned. Others just want to know.