HUNTINGTON'S DISEASE

Patient Information Fact Sheet

What is Huntington's disease?

Huntington's disease, which is often called HD, is a hereditary disorder of the central nervous system. It used to be known as Huntington's Chorea or HC. Huntington's disease usually develops in adulthood and can cause a very wide range of symptoms—including physical, mental and emotional symptoms.

What causes Huntington's disease?

Huntington's disease is caused by a faulty gene on chromosome 4. The gene, which produces a protein called huntingtin, was discovered in 1993. Though the mechanism is not yet understood, the faulty gene leads to damage of the nerve cells in areas of the brain including the basal ganglia and cerebral cortex. This leads to gradual physical, mental and emotional changes. A person with a parent who has Huntington's disease is born with a 50–50 chance of inheriting the faulty gene. Anyone who inherits the faulty gene will, at some point in their life, develop the disease. A genetic test is available from Regional Genetic Clinics throughout the country. This will usually be able to show whether someone has inherited the faulty gene, but it will not show the age at which they will develop the disease.

What are the early symptoms of Huntington's disease?

The symptoms of Huntington's disease usually develop when people are between 30 and 50 years old, although they can start much earlier or much later and can differ from person to person, even within the same family. Sometimes, the symptoms are present for a long time before a diagnosis of Huntington's disease is made. This is especially true when people are not aware that Huntington's disease runs in their family.

The early symptoms include slight, uncontrollable muscular movements; stumbling and clumsiness; lack of concentration and short-term memory lapses; depression and changes of mood, sometimes including aggressive or antisocial behavior. Great strain is put on relationships if unexpected temper outbursts are directed toward a person's partner or family. The time before a diagnosis is made can be very confusing and frightening because people do not understand what is happening and why. Some people who know they are at risk anticipate the first signs that they are developing the disease. They may worry about simple things such as dropping a cup, forgetting a name or becoming unusually bad-tempered. Most people do these things occasionally—whether they are at risk from Huntington's disease or not—so they could be worrying unnecessarily. Anyone who is concerned should speak to their doctor who may refer them to a neurologist for tests. These tests could include a number of simple assessments and possibly a brain scan. The genetic test referred to earlier may also be used to aid diagnosis.

How does Huntington's disease progress?

As the disease progresses, a person may experience many different symptoms, including involuntary movements, difficulty in speech and swallowing, weight loss, emotional changes resulting in stubbornness, frustration, mood swings and depression. Cognitive changes that people experience result in a loss of drive, initiative and organizational skills, which may lead others (who are not aware

of the disease) to see the person as lazy. There may be difficulty in concentrating on more than one thing at a time. Sometimes it is the psychological problems, rather than the physical deterioration, that causes more difficulty for the person with Huntington's disease and their career.

Some changes that are definitely attributable to the disease may be exacerbated by other factors, such as depression, anxiety and frustration. It is depressing to have a serious illness and extremely frustrating not to be able to do things that previously seemed commonplace. In the later stages of the disease, full nursing care will be needed. Secondary illnesses, such as pneumonia, are often the actual cause of death.

How is Huntington's disease treated?

Currently, there is no cure for the illness but there are many ways to manage symptoms effectively. 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 improve symptoms such as involuntary movements or behavioral problems. Social services in your local area can assist with practical issues such as appropriate adaptations to your home if necessary and also assist with care at home or respite care. They can also assist with the provision of equipment if necessary. A referral can be made through your doctor.

Further information

National Institute of Neurological Disorders and Stroke:
www.ninds.nih.gov/disorders/huntington/huntington.htm

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