FACT SHEET

Understanding Huntington Disease

THIS FACT SHEET describes the nature of Huntington disease, its cause and who develops it. It covers symptoms, diagnosis, genetic testing, the effects over time, appropriate therapies and care-giving.

What is Huntington Disease?

Huntington Disease (HD) is a genetic neurodegenerative condition, which means it affects the brain and is inherited through a family line. HD destroys the brain cells that affect our emotions, intellect and movement. People with HD experience uncontrollable jerking movements (chorea) of the limbs, torso and face, loss of mental abilities and may experience behavioural and personality changes.

What Causes HD?

HD is caused by a faulty gene that is passed on by one parent. Researchers do not yet understand how, but the faulty gene leads to damage of nerve cells (neurones) in the brain. Two areas of the brain in particular are affected: the basal ganglia, which controls and coordinates body movements and the cerebral cortex, which is responsible for processes such as thought, emotions, perception and planning.

Who Develops HD?

About 5 – 10 people per 100,000 will develop Huntington Disease.

A person who has the faulty HD gene has a 50% chance of passing it on to each of their children. If the child inherits the gene, HD symptoms will develop.

Genetic testing: People at risk of inheriting the faulty HD gene can have genetic testing done to determine their status. Testing is only available to people aged 18 years and over. A doctor will first examine you for symptoms of HD. Testing is available at the Murdoch Institute, which is located at the Royal Children's Hospital. Costs are covered by Medicare.

Counselling: Because the results of genetic testing can be upsetting and create difficult decisions, anyone who chooses to have it must first see a counsellor. Counsellors discuss the possible outcomes and consequences of genetic testing thoroughly with a person to help them determine if it is a wise option.

To speak to a support worker, at the Australian Huntington's Disease Association (Vic) Inc: telephone (03) 9563 3922.

To speak to a counsellor, call Genetic Health Services Victoria: telephone (03) 8341 6201.

Symptoms

Symptoms are progressive – they become worse over time. They usually appear between the ages of 35 and 45, occasionally earlier and sometimes later. HD affects each person differently but symptoms fall into three main categories:

- physical
- cognitive
- emotional/behavioural

Physical: Physical symptoms may start with mild twitching of the fingers, toes, face and limbs. The person may feel less coordinated and fumble or knock things over. They may trip and bump into

things. Twitching progresses to jerky movements of the arms and legs, known as chorea (a small number of people do not experience this). Over time, speech becomes slurred and swallowing becomes difficult.

Cognitive: The term "cognitive" refers to activities such as thinking, planning, organising and our ability to judge and perceive things. Short term memory problems may become evident, for instance a person may forget to turn off the stove. Tasks that require concentration may become more difficult, such as mental arithmetic, planning a work schedule, making decisions or reading. But people with HD do not lose their ability to recognise the people around them. They will always be able to recognise people and know where they are.

Emotional/behavioural: In the early stages, a person may be more irritable and moody. Depression affects about one in three people. Gradually the person may become apathetic or aggressive and develop inappropriate behaviours, such as laughing or crying for no apparent reason. Changes of this nature are often an exaggeration of an existing personality trait - a highly strung person becomes more so.

Diagnosis

Diagnosis of HD is usually made by a neurologist (brain specialist) or a psychiatrist and is based on a family history of HD, cognitive and physical symptoms. Genetic testing may also be used.

Sometimes people who know they are at risk of inheriting HD exhibit symptoms long before they are ready to seek a diagnosis. It's important for carers to understand this reluctance to accept the possibility of having the condition and to remain as supportive as possible. Family members can speak to the person's doctor about their concerns and keep in touch with the Australian Huntington's Disease Association (AHDA) (Vic.) Inc. for support.

Treatment

There is currently no cure for HD but there are many ways to improve a person's quality of life and cope with the symptoms.

What is the Outlook?

HD progresses slowly and a person may live for 15 to 25 years after developing their first symptoms. Generally, the older the age of onset, the more slowly the condition progresses.

Managing HD

Most people with HD see a general practitioner (GP) for their day-to-day medical care and have periodic appointments with a specialist, such as a neurologist or a psychiatrist. Because HD is a relatively rare condition however, it's not uncommon for GPs and other therapists to know little about it. The AHDA (Vic.) Inc. has a doctors' information kit and can help with contacts for HD-aware therapists and clinics.

Medical treatment: Medications are available to help lessen the involuntary movements but these often have side effects. It is best to discuss the options carefully with your GP or specialist. Depression is common in HD but this can be treated with medication.

Supporting therapies: Various physical therapists will be involved in the long-term care of someone with HD. The earlier you begin therapy with these specialists, the better. The AHDA (Vic.) Inc. can help you locate appropriate therapists.

For example, a physiotherapist helps a person maintain their physical skills for as long as possible, then teaches them new ways of doing things.

An occupational therapist can help to make their home safe and easy to manage and assist with a range of practical difficulties that arise in everyday living (See Fact Sheet 13: Adapting Your Home).

Speech pathologists can help with strategies and devices to aid speech and communication as these become more difficult. (See Fact Sheet 7: Speech and Communication Problems).

A neuropsychologist can provide memory and behaviour-management strategies.

Nutrition: Establishing good nutrition is particularly important for people with HD. As the disease progresses, swallowing becomes difficult

and weight loss is a common problem. Meal times can become unpleasant and even frightening for the person and their carer. It's wise to engage a speech pathologist early on to learn strategies to make swallowing as safe as possible. (See Fact Sheet 8: Eating and Swallowing Problems.)

Equally, a dietician can devise meal programs that are enjoyable, high in nutrition and easy to manage.

Caring for Someone with HD

Staying active: A person can slow the progression of HD by staying physically, mentally and socially active for as long as possible. Carers can encourage their family member or friend to do things they have enjoyed in the past, such as walking, reading, listening to news, going to the football or a movie or having coffee with friends. People with HD often have difficulty organising themselves and getting started. Carers can help a great deal by kick-starting activities. For example, don't just suggest the person join the gym, go with them and help them organise a schedule for visits.

Counselling and support: The AHDA (Vic.) Inc. has a team of support workers who well understand the problems and stresses that people with HD and their families encounter. Do make good use of these services. We can also put you in touch with support systems within your own community.

Contact

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