What is Huntington’s Disease?

Huntington’s Disease Association of Ireland - Supporting people impacted by HD
WHAT IS HUNTINGTON’S DISEASE?

Huntington’s Disease (HD) is a hereditary neurodegenerative illness with physical, cognitive and emotional symptoms.

HD is caused by an alteration in the ‘huntingtin gene’ (HD gene), on chromosome 4. The alteration or gene change causes cells to create a harmful protein called mutant huntingtin.

The harmful protein behaves differently from the normal protein and causes cells in certain parts of the brain to malfunction and die. As brain cells die, changes to movement, thinking and emotion occur and gradually worsen over time.

Symptoms can begin at any age; but for most people with HD symptoms usually start between 35 and 50 years of age. About 5 -10% of people with HD have symptoms before age 20 (Juvenile HD) and 10% have onset after age 60 (Late onset).

HOW IS HD INHERITED?

Huntington’s Disease is a genetic condition and the HD gene is dominant. This means that if either parent has the altered HD gene, each child is said to be at risk and has a 50% chance of inheriting or not inheriting Huntington’s disease. Males and females have the same risk of inheritance.

If you have the altered HD gene you will almost always develop the illness. If you are at-risk and do not inherit the altered HD gene, you will not develop the illness and you cannot pass it on to your children.
Some people develop HD without ever knowing they were at risk, because they have no known family members with the condition. This may be explained by a parent’s misdiagnosis, the early death of a parent before symptoms began, by adoption, or by mistaken paternity. Others represent ‘new mutations’, caused by rare expansions of parental genes with a high-normal CAG repeat number (27-35 repeats) into the affected range in the child. A genetic counsellor can provide information on this.

Symptoms vary from person to person and at different stages of the illness. You may start to notice subtle changes in the way you move, how you think and how you feel.

HD usually progresses slowly over a period of 15-25 years resulting in gradual physical, cognitive and emotional changes which may include:

- **Physical symptoms**: weight loss, involuntary movements (chorea), diminished coordination, difficulty with walking, talking and swallowing
- **Cognitive symptoms**: impaired insight, problems with focusing, planning, recalling information and making decisions
- **Emotional symptoms**: depression, apathy, irritability, anxiety, obsessive behaviour
WHAT TREATMENTS ARE AVAILABLE?

Clinical assessment of motor, cognitive and mental health symptoms should determine services required.

While there is no cure for HD at this time, medical intervention including rehabilitative therapies such as physiotherapy, occupational therapy, speech and language therapy and diet and nutrition expertise can help manage symptoms.

Support and information, neuropsychological and psychiatric services can help with cognitive and neuropsychiatric changes.

A healthy diet, regular exercise, keeping our brain active and avoiding undue stress are important. This self-care can help people with HD cope with the symptoms of the illness and maintain a better quality of life for many years.

Keeping a healthy body weight is essential for managing physical health and well-being. Anecdotal evidence suggests that this can help to reduce involuntary movements, and improve speech, swallowing and mood.

People with HD who are under weight or dropping weight should seek a referral to a dietician and speech and language therapist for advice.

The European HD Network (EHDN) in collaboration with the journal *Neurodegenerative Disease Management* have published a set of ‘open access’ HD guidelines. The guidelines include advice on physiotherapy, nutrition, speech and communication, and occupational therapy.

See [www.huntingtons.ie](http://www.huntingtons.ie)
HOW MANY PEOPLE HAVE HD?

Professor Patrick Morrison conducted a survey in Northern Ireland in 2001 which estimated a prevalence of 10-16 people per 100,000 (Lancet Neurology).

His analysis of more recent data suggests a prevalence of 14-16 per 100,000. Based on the Republic of Ireland population of 4.773 million (2016), this would indicate more than 700 people with HD in Ireland and a further 3,000 at risk.

The secrecy and stigma previously attached to HD is thought to have influenced the under-reporting of the illness. This highlights the need to raise awareness and encourage affected families to access support so that Health Services can provide adequate care for the actual number of people impacted by Huntington’s Disease.

HOW IS HD DIAGNOSED?

The discovery of the HD gene in 1993 resulted in the availability of an accurate blood test.

If you are worried that you have symptoms, you can ask your GP for a referral to a neuroligist.

A diagnostic test is used to confirm the diagnosis of HD in a person showing symptoms of the disease.

Special consideration should be made regarding the effects of an individual’s diagnostic test results on their family.
IS THERE A TEST FOR PEOPLE AT RISK?

A gene test is available which allows a person at risk to find out if they carry the altered HD gene and will someday develop the illness. The predictive test cannot determine when the symptoms of HD will begin or how severe the symptoms will be. Not everyone with a family history of HD wishes to take a gene test. Taking the test remains an individual choice.

Details on genetic counselling and testing are available from:

The Department of Clinical Genetics, Our Lady's Children's Hospital, Crumlin, Dublin 12.
Tel: (01) 409 6739.
Web: http://www.olchc.ie/

ADJUSTING TO A DIAGNOSIS

A diagnosis of HD in a family brings with it a range of emotions. Fear, anger, shock, denial, loss are all normal responses, and these reactions are part of the journey in coming to terms with the diagnosis.

Some families may already be familiar with HD because they have relatives with the condition. Others may have never heard of HD and may be shocked by the diagnosis. Getting support and information is the first stage in helping to adjust.

HD is often referred to as a family disease and it is important to recognise that each family member, whether they have HD or not, will be impacted by HD. People who get a favourable gene test result often experience survivor guilt. Many family members become carers for relatives who have HD.
DELAYING ONSET

Research suggests that by leading a mentally and physically healthy and active lifestyle, you can delay the onset of symptoms. Keeping physically and mentally active, maintaining a healthy diet, enjoying life and avoiding excessive stress will have a positive impact for people at risk.

RESEARCH

Significant progress has been made by researchers world-wide to understand HD and to develop potential therapies to halt or reverse the disease. There is now greater hope than ever before that therapies will be found to fight this disease. Contact HDAI for information.

HUNTINGTON’S DISEASE ASSOCIATION OF IRELAND (HDAI)

HDAI is a voluntary organisation which provides support, information and advocacy for individuals and families living with Huntington’s disease. HDAI aims to raise awareness and understanding of HD with health and social care professionals.

Services include:

- National Information & Support Service
- Family Support Officer
- Access to counselling
- Regional support groups
- Carers workshops
- Information seminars
- Annual respite weekend
- Publications
- Newsletters
Huntington’s Disease Association of Ireland, Bank of Ireland, Smithfield, Dublin 7.
Account No. 54757711
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Donate online via www.huntingtons.ie

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