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# Fact Sheet

## Huntington's Disease

### Definition

Huntington's Disease (HD), also called Huntington's chorea, is an inherited brain disorder that results in loss of physical control and mental capacity. Symptoms typically appear between the ages of 30 and 50, but may appear earlier or later. HD is characterized by progressive physical, cognitive and psychological deterioration.

### What happens when you have HD?

Genes are made up of long strands of building blocks called *nucleotides*. In an individual with HD, there is a surplus of a specific building block—a genetic stutter (a stretch of DNA repeated over and over at one end of a gene) on chromosome four. This eventually leads to the symptoms of HD. Since the genetic problem causes nerve cell loss in the brain, specifically the basal ganglia, symptoms of HD may develop gradually, and will affect an individual's ability to move, think and process thoughts and feelings. The progression of HD is frequently divided into stages, and each stage marks a loss in ability, or a change in situation. As HD progresses, additional neurons are damaged in the brain, leading to further physical, cognitive and psychological decline.

### Facts

Though once considered a rare disease, HD is one of the more frequently encountered hereditary diseases. Nationwide, an estimated 30,000 people have Huntington's Disease. Because individuals affected by HD need extensive care from their families and loved ones, this devastating condition has an effect on a wide circle of people close to those who are ill.

HD affects both women and men and all ethnic groups. People from two to 80+ years of age can develop HD. The disease has a gradual decline over a period of 10 - 25 years, typically leading to complete dependence on others. The affected person eventually dies from complications, such as heart failure or aspiration pneumonia.

Approximately 200,000 Americans are at risk of inheriting the disease from an affected parent. Those who have a parent with HD have a 50 percent (i.e., one in two) chance of inheriting the defective gene. In other words, passing the HD gene to a child is random; some parents may not pass the gene to any of their children; some pass it to all of them. HD does not skip generations. Therefore, if a child does not inherit the gene, they cannot pass it on to future generations. Every person who inherits the HD gene will eventually develop the disease if they don't die of other causes (stroke, cancer, etc.) before HD progresses.

### Symptoms

An individual with Huntington's Disease often has problems in three areas: control of body movement, cognitive (or mental) functioning and psychological issues. The intensity and number of symptoms vary case by case. Symptoms experienced by those with HD may include:

- **Movement** - Quick, dance-like, uncontrollable movements of the limbs are typical of those with HD, and are commonly called chorea by health professionals. Movements of the torso and face are the most obvious. All movement disorders

tend to progress over time, both in frequency and severity. Movements may be grossly exaggerated, causing what was intended to be a small movement to become tremendous instead. For example, an intended small shift in bed may cause the individual to actually vault out of bed. Eventually rigidity or muscle stiffness may occur. HD may affect your loved one's ability to walk—he or she may become unsteady, or even appear “drunk.” In later stages of the disease, swallowing becomes increasingly difficult (dysphagia), and choking is a major concern.

- **Cognitive** - Huntington's Disease causes changes in the brain that slow processing of information and hamper organizational ability. Thus, your loved one may find it increasingly challenging to organize typical household matters, cope with new situations, or switch from task to task (multi-tasking). Because individuals with HD have difficulty processing information, they may have difficulty remembering words, and speech may become challenging. Individuals with HD may exhibit poor judgment and problems with short-term memory. Declining cognitive function is common as the disease progresses, although those with HD continue to recognize their caregivers, are aware of what is happening around them, and can give feedback about their condition and care.
- **Psychological** - Some people with HD may have difficulty controlling impulses and emotions, resulting in outbursts, yelling or aggression. Mild to severe depression, common in most individuals with chronic, long term conditions, is one of the primary complaints of HD. Other psychological symptoms include

irritability and anxiety. Others may exhibit schizophrenic-like behavior, such as hallucinations, mania, anxiety and psychosis.

## **Diagnosis of Huntington's Disease**

A thorough neurological exam that uses the Unified Huntington's Disease Rating Scale (UHDRS) and an extensive patient and family medical history can confirm the presence of HD. Symptoms exhibited, however, may or may not be caused by the disease. A physician or neurologist, preferably one familiar with HD, can help to make this determination. Genetic testing is the most conclusive and definitive way to determine if your loved one will develop, or has developed, Huntington's Disease.

## **The HD Gene and Genetic Testing**

In 1993, the discovery of the gene that causes Huntington's Disease led to the development of a direct gene test for HD. This blood test, also called a presymptomatic test, can be used to predict Huntington's Disease before symptoms develop. The test consists of a series of visits to a genetic testing center for genetic counseling, a thorough neurologic exam, a psychological interview, discussion of positive or negative results, and follow-up. Test results cannot indicate *when* symptoms will appear, although research is ongoing in this area.

A second blood test, called a confirmatory test, confirms the presence of the HD gene. The procedure for this test is the same as the presymptomatic test; however, the test is given after symptoms of the disease have begun to appear. Prenatal testing is available to determine if a fetus carries the HD gene. Due to the potential burden of the knowledge that one will develop this chronic disease, however, it is recommended that children not be tested until they are at least 18 years of age. In the absence of a cure, at-risk individuals may prefer to live with uncertainty, rather than take the test. The decision is a

personal one and should be discussed with a genetic counselor, families and loved ones.

Finding a testing center that follows the Huntington's Disease Society of America (HDSA) guidelines for genetic testing is very important. HDSA publishes an up-to-date listing of these testing centers (see Resources).

## **Research**

Research is ongoing to develop new strategies to fight HD. Researchers are currently studying methods to delay the onset of HD or to halt the progression while a cure is developed. Other research efforts with animal models are determining the effects of HD, how it manifests, and if or how successful animal trials can be translated to treatments that work in humans. Research on the effects of surgical transplantation of stem cells and tissue transplantation is also underway. Both basic (laboratory) and clinical (testing of medications and treatments) research continue to facilitate new drug testing. To stay informed of, or participate in ongoing clinical trials, contact your local chapter or the national office of HDSA.

## **Treatment**

Currently, no medications can halt or slow the progression of HD. Thus, symptom management is the goal of treatment for HD. Medication may reduce involuntary movements and emotional disorders for some HD patients. Neuroleptic drugs are typically prescribed to reduce involuntary movement. Anti-depressants or anti-psychotic medications are generally recommended for psychological symptoms. Families should proceed with caution when using new medications, however, as some individuals with HD may be more sensitive to side effects than others. It is important to document side effects of new medications or changes in behavior after a medication has been added to your loved one's routine.

Many other treatments can assist those with HD and their loved ones. Physical therapists can assist with early balance and walking problems and provide exercises to build strength. Exercise can also help combat depression. Occupational therapists can provide strategies to make your home safer and more livable for your loved one with HD, improve communication, and suggest ideas to offset cognitive decline.

Some helpful techniques that have worked for caregivers include labeling items around the house (for example, to indicate the contents of a kitchen cabinet), using lists and notes, and communicating with short, direct instructions. HD affects metabolism, and those with this disease may burn calories at a much higher than average rate. Thus, it may be important to keep your loved one on a high calorie diet. Dieticians can assist with diet changes and meal planning. Since the ability to communicate continually declines for those who have HD, a speech pathologist may also be particularly helpful at all stages of the disease. Ideally, treatments should be coordinated, and medications should be prescribed by health professionals who are thoroughly familiar with HD.

## **The Role of Health Professionals**

When your loved one is being tested or receiving treatment for HD, you may seek the expertise of many different health professionals. People who have HD experience a number of symptoms and potentially many stages, so you may interact with what might seem like a great many physicians, social workers, physical therapists and other health care providers. It can be overwhelming to coordinate care and to find appropriate resources and professionals who are trained in HD care. Keeping a notebook or "health journal" of the interactions with health care providers that you see, treatments administered, and medications prescribed will help to keep appointments organized and will provide a personal record of care.

By seeking early intervention from health professionals such as physical, occupational and speech-language therapists, those with HD can have more control over the disease and over their well-being.

## **Getting Support**

Due to the strong emotional impact of the diagnosis of this chronic, hereditary condition, and the stress of knowing that others in the family may be at risk, participation in support groups can be very helpful. Support groups provide a safe, caring environment in which to share information about individual experiences and challenges of HD with others in similar situations. Topics discussed may include adjustment, coping, disease course, family issues, frustrations, insurance, medication and more. Both online and in-person support groups are available for individuals with HD, their family caregivers, and for those living at risk. Counseling also may be helpful for individuals, couples, or families who are learning to cope with chronic illness or periodic health crises.

Family and friends who provide assistance to a loved one with HD may be at increased risk of poor health, depression and isolation. It is important for the health of the family caregiver, as well as the health of the person with HD, for their caregivers to take some time off from caregiving (known as respite), get enough sleep and have a support system of their own. Respite care can be arranged with the help of family members, friends, volunteer services, independent living centers, social service organizations or home care agencies.

Caregivers who make time for themselves can give better care to their loved ones. For more information, see Family Caregiver Alliance's Fact Sheet, [Taking Care of YOU: Self-Care for Family Caregivers](#).

Today, more than ever, there is hope for people with HD. Rapid progress is being made toward potential treatments. Likewise, quality of care for people with HD

has improved greatly in recent years. By learning strategies to help them cope with HD, people can live meaningful, productive lives well into the final stages.

## **Recommended Readings**

***A Caregiver's Handbook for Advanced-Stage Huntington's Disease***, Jim Pollard, 2000, Huntington's Disease Society of America (HDSA), New York, NY.

***A Physician's Guide to the Management of Huntington's Disease***, (2<sup>nd</sup> edition) Neal Ranen, Carol Peyser, Susan Folstein, 1999, Huntington's Disease Society of America (HDSA), New York, NY.

***Huntington's Disease: The Facts***, Oliver Quarrell, 1999, Oxford University Press.

***Juvenile HD: A Guide for Families***, 2001, HDSA, New York, NY.

***Long Term Care: A Guide for Families***, 2001, HDSA, New York, NY.

***Understanding Behavior in Huntington's Disease: A practical guide for individuals, families and professionals coping with HD***, Jane S. Paulsen, 2000, HDSA, New York, NY.

***Walking the Tightrope: Living at risk for Huntington's Disease***, Randi Jones, 1998, Huntington's Disease Society of America, 1998.

***Fact Sheet: Taking Care of YOU: Self-Care for Family Caregivers***, Family Caregiver Alliance. [taking-care-you-self-care-family-caregivers](#)

## Credits

HOPES, *The Diagnosis of Huntington's Disease*, 2003, [www.stanford.edu/group/hopes](http://www.stanford.edu/group/hopes).

Huntington's Disease Society of America, *Huntington's Disease*, 2003, [www.hdsa.org](http://www.hdsa.org).

Huntington's Disease Society of America, *Physical and Occupational Therapy for Huntington's Disease*, 2003, [www.hdsa.org](http://www.hdsa.org).

Huntington's Disease Society of America, *Genetic Testing*, 2003, [www.hdsa.org](http://www.hdsa.org).

Huntington's Disease Study Group, *What is Huntington's Disease?*, 2002, [www.huntington-study-group.org](http://www.huntington-study-group.org).

## Resources

### Family Caregiver Alliance

785 Market Street, Suite 750

San Francisco, CA 94103

(415) 434-3388

(800) 445-8106

Web Site: [caregiver.org](http://caregiver.org)

E-mail: [info@caregiver.org](mailto:info@caregiver.org)

Family Caregiver Alliance (FCA) seeks to improve the quality of life for caregivers through education, services, research and advocacy.

FCA's National Center on Caregiving offers information on current social, public policy and caregiving issues and provides assistance in the development of public and private programs for caregivers.

For residents of the greater San Francisco Bay Area, FCA provides direct family support services for caregivers of those with Huntington's, Alzheimer's disease, stroke, ALS, head injury, Parkinson's and other debilitating disorders that strike adults.

## Huntington's Disease Society of America

158 West 29th Street, 7th Floor

New York, N.Y. 10001-5300

(800) 345-HDSA

(212) 239-3430 (fax)

[www.hdsa.org](http://www.hdsa.org)

The Huntington's Disease Society of America (HDSA) is dedicated to finding a cure for Huntington's Disease through the promotion and support of both clinical and basic research, while also providing support and services for those living with HD and their families. Through its nationwide network of volunteer-based chapters and affiliates, HDSA provides family services, support groups, educational programs and regional referrals. HDSA Centers of Excellence assist families in managing the challenges of HD, as well as providing vital information about care, financial planning and legal issues. Contact HDSA to obtain a current publications list which includes written materials and audiovisuals pertaining to all aspects of Huntington's Disease.

### International Huntington Association

Callunahof 8

7217 ST Harfsen

The Netherlands

Tel: 31 - 573 - 431 595

Fax: 31 - 573 - 431 719

[www.huntington-assoc.com](http://www.huntington-assoc.com)

### American Speech Language Hearing Association

10801 Rockville Pike

Rockville, MD 20852

[www.asha.org](http://www.asha.org)

**National Institute of Neurological Disorders and Stroke**

P.O. Box 5801  
Bethesda, MD 20824  
(800) 352-9424 or (301) 496-5751  
TTY (for people using adaptive equipment): (301) 468-5981  
[www.ninds.nih.gov](http://www.ninds.nih.gov)

**National Society of Genetic Counselors**

233 Canterbury Dr.  
Wallingford, PA 19086-6617  
(610) 872-7608  
[www.nsgc.org](http://www.nsgc.org)

The National Society of Genetic Counselors (NSGC) can assist individuals and their families in locating a genetic counselor. NSGC does not, however, maintain information on specific genetic disorders.

**Helpful Websites**

**Family Caregiver Alliance's  
Huntington's Disease and Movement Disorders  
Online Support Group**  
[caregiver.org](http://caregiver.org)

**Huntington's Disease Society of America**  
[www.hdsa.org](http://www.hdsa.org)

**HD Advocacy Center**  
[www.hdac.org](http://www.hdac.org)

**HOPES: Huntington's Outreach Project for  
Education, at Stanford**  
[www.stanford.edu/group/hopes](http://www.stanford.edu/group/hopes)

For local resources:

The Huntington's Disease Support Information website has an extensive listing of HD state resources.

<http://www.hdsa.org/sg>

**Huntington Study Group**

[www.huntington-study-group.org](http://www.huntington-study-group.org)

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**Related Health or Illness Resources**

- [AoA Announces Availability of Approximately \\$2.25 Million for Lifespan Respite Care Programs](#)
- [Huntington's Disease & Movement Disorders Discussion Group](#)
- [New Documentary on Huntington's Disease: 'The Lion's Mouth Opens'](#)
- [With Far Fewer Benefits I Could've Kept Him Home](#)