

# Huntington's Disease



## A Family Guide



Huntington's Disease  
Society of America

# Mission

To improve the lives of everyone affected by Huntington's disease and their families.

# Vision

A world free of Huntington's disease.

© 2016, Huntington's Disease Society of America. All Rights Reserved.  
Printed in the United States.

No portion of this publication may be reproduced in any way without the expressed written permission of HDSA.



Huntington's Disease  
Society of America

# Huntington's Disease

## Family Guide Series

Kathleen Shannon, M.D.  
Rush University Medical Center  
Medical Editor

Debra Lovecky  
Director of Educational Programs, HDSA  
Editor



## About the Huntington's Disease Society of America (HDSA)

---

The Huntington's Disease Society of America is the premier nonprofit organization dedicated to improving the lives of everyone affected by Huntington's disease. From community services and education to advocacy and research, HDSA is the world's leader in providing **help for today, hope for tomorrow** for people with Huntington's disease and their families.

The Huntington's Disease Society of America was founded in 1968 by Marjorie Guthrie, the wife of music icon Woody Guthrie. Woody died from HD complications in 1967 when he was only 55 years old, but the Guthrie family legacy lives on at HDSA to this day.

HDSA Currently Supports:

- HDSA Centers of Excellence
- Volunteer-led Chapters & Affiliates across the United States
- HD Research Initiatives
- HDSA HD TrialFinder clinical trial matching service ([www.HDTrialfinder.org](http://www.HDTrialfinder.org))
- Social Workers
- Support Groups
- HD Advocacy Programs
- And Much More!

For more information on how to get involved or for more assistance please visit [HDSA.org](http://HDSA.org) or call (800) 345-HDSA.

**In the battle against Huntington's disease  
no one fights alone.  
At HDSA, family is everything.**

### Disclaimer

Statements and opinions in this book are not necessarily those of the Huntington's Disease Society of America (HDSA) nor does HDSA promote, endorse, or recommend any treatment or therapy mentioned therein. Nothing in this presentation claims to diagnose any condition, prescribe any specific treatment, or to take the place of consultation with qualified professionals.

# Contents

---

Overview .....	4-8
What is HD? .....	5
Inheritance .....	6
Juvenile Onset Huntington's Disease (JHD) ...	6
Clinical Diagnosis .....	7
Stages of HD .....	7
Living with HD .....	9-13
The Three Spheres of HD .....	9
• Movement Disorders .....	10
-Chorea .....	10
-Treating chorea .....	10
-Impairment of voluntary movements .	11
-Treatment.....	12
• Cognitive Disorders.....	12
• Emotional/Behavioral Disorders.....	13
-Treatment.....	13
Being at-risk for HD .....	14
Genetic Testing .....	14
The Search for Effective Treatments and a Cure .....	16

# Overview

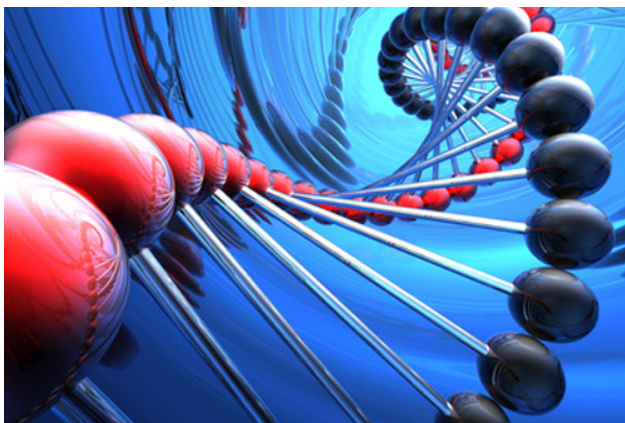
---

Huntington's disease is a fatal genetic disorder that causes the progressive breakdown of nerve cells in the brain. It deteriorates a person's physical and mental abilities usually during their prime working years and has no cure.

Every child of a parent with HD has a 50/50 chance of carrying the faulty gene that causes Huntington's disease. Today, there are 30,000 symptomatic Americans and more than 200,000 at-risk of inheriting the disease.

In approximately 10% of cases, Juvenile Huntington's disease (JHD) affects children or adolescents and JHD typically progresses more rapidly than adult onset HD.

The symptoms of HD are described as having ALS, Parkinson's and Alzheimer's diseases – ***simultaneously***.



HD affects persons of all races, genders and ethnic groups around the world. It takes its name from Dr. George Huntington, a Long Island, NY physician who described what he called "hereditary chorea" in 1872. Chorea, from the Greek word for dance, refers to the involuntary movements that are a common symptom of HD.

# What is HD?

---

HD is a “neurodegenerative disease,” which means that there is a progressive death of nerve cells in the brain. Symptoms typically appear in mid-life, between the ages of 30 and 50, and progress for 10-25 years. However, the disease can strike young children, adolescents and the elderly.



HD is a complicated disease that affects the body, the mind and the emotions. There are symptoms of HD that are easy to see, like chorea/involuntary movements, and there are those that are less visible, such as slowed thinking, impulsiveness, or depression. Symptoms of HD vary widely from person to person, even within the same family. In addition, symptoms change over time as the disease progresses. Symptoms of HD and their effects are discussed in detail in the “Three Spheres of HD” section beginning on page 9.

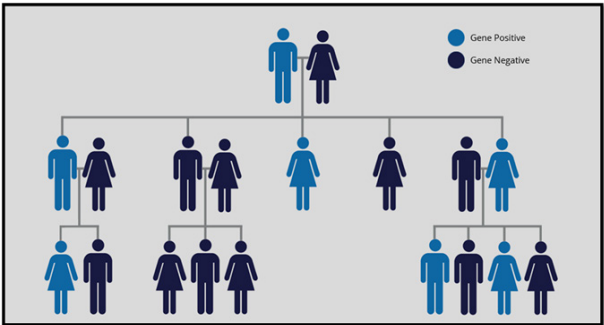
Symptoms appear gradually and a person with HD can maintain his or her independence for many years. As you will learn, HD affects all aspects of a person’s life and treating it requires a broad-based approach. Prolonging independence may call for a team of health care professionals that may include a primary care physician, neurologist, social worker, speech-language pathologist, nutritionist, occupational therapist and physical therapist, among others. Referrals to professionals who understand HD can be provided by the HDSA national office, your local HDSA chapter or by visiting [www.hdsa.org](http://www.hdsa.org).

# Inheritance

---

HD is known as the quintessential family disease. Though everyone is born with the huntingtin gene, the disease is caused by a genetic variant that is passed from parent to child. It is not contagious in any way. Only a person who inherits this genetic variant will develop the illness or pass it on to their children. Every person who carries the genetic variant will eventually develop symptoms, if they live long enough.

Every child born to an affected parent has a 50% chance of inheriting the genetic variant that causes the disease. Males and females are equally affected. Those who have not inherited the genetic variant will not get the disease and they cannot pass it on to their children. HD does not “skip” generations.



## Juvenile Onset Huntington's Disease (JHD)

---

In approximately 10% of cases, HD affects children or adolescents. The symptoms of JHD are somewhat different than adult HD and may include stiff or awkward walking, increased clumsiness or changes in speech. The ability to learn new information may decline and the child may lose skills they previously had. JHD typically progresses more rapidly than adult HD. A variety of materials on JHD are available from the HDSA website, [www.hdsa.org](http://www.hdsa.org), or by calling 800-345-HDSA.



# Clinical Diagnosis

---

Although the genetic variant is present from birth, a clinical diagnosis of HD indicates that symptoms have started. A diagnosis of HD can only be accomplished by conducting a comprehensive neurological evaluation that is best performed by a specialist in HD or in movement disorders. A genetic test may be used to help confirm or rule out a diagnosis; however, a positive test result, indicating the presence of the variant, is not enough by itself to confirm a diagnosis of clinical HD.

For some, a diagnosis can be a relief, providing an explanation for the changes in movements, thinking and emotions. Others find the news very upsetting. A state of “denial” is not uncommon when a diagnosis of HD is first made. Whatever the reaction, it is helpful to talk about the situation with a social worker, genetic counselor or therapist.

## Stages of HD

---

Although symptoms of HD vary from person to person, even within the same family, the progression of the disease can be roughly divided into three stages.

*Early stage HD* usually includes subtle changes in coordination, perhaps some involuntary movements (chorea), difficulty problem solving and often a depressed or irritable mood. Medications can be helpful in treating depression or other emotional problems. The effects of the disease may affect the person’s ability to work at their customary level or be less functional in their regular activities at home.

In the *middle stage*, the movement disorder may become more pronounced. Medication for chorea may be considered to provide relief from involuntary movements.

Occupational and physical therapy may be needed to help the person maintain control of voluntary movements and to deal with changes in thinking and reasoning abilities. Diminished speech and difficulty swallowing may require help from a speech-language pathologist. Ordinary activities will become harder to manage.

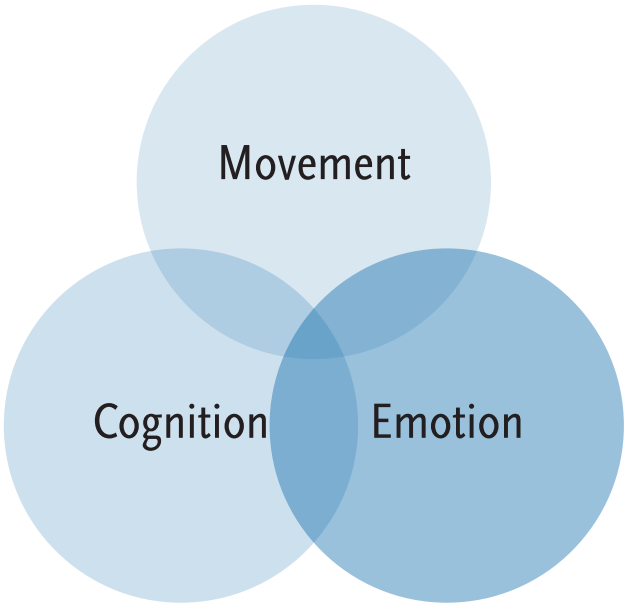


In the *late stage*, the person with HD is totally dependent on others for their care. Choking becomes a major concern. Chorea may be severe or it may cease. At this stage, the person with HD can no longer walk and will be unable to speak. However, he or she is generally still able to comprehend language and retains an awareness of family and friends. When a person with HD dies, it is typically from complications of the disease, such as choking or infection and not from the disease itself.

In all stages of HD, weight loss can be an important complication that can correspond with worsening symptoms and should be countered by adjusting the diet and maintaining appetite. Consultation with a dietitian can help.

# Living with HD

## The Three Spheres of HD



Understanding the wide-ranging impact of HD on a person can help family and friends remain supportive as he or she faces the challenges of the disease. Over time, HD affects all spheres of a person's life. It causes changes in the brain that impact movement, cognition (thinking), emotions and behavior, and changes in one area often affect another.

## Movement Disorders

Abnormal movements are the most *visible* symptoms of HD and can include clumsiness, loss of balance and fidgeting. Difficulties with movements may include quick involuntary movements, known as chorea, twisting postures (dystonia), and reduced speed and accuracy of fine movements. The movement disorders of HD often increase with stress or excitement.

### Chorea

Adults with HD often appear restless and make “*fidgety*” gestures or grimaces. These involuntary movements are known as chorea. Chorea can be mild or severe and is often the first *physical* symptom of HD. It may appear as random, jerky movements of the fingers and toes. Later, the person may develop a distinctive “scissoring” or lurching walk. Over time, involuntary movements of the head, trunk and limbs may become severe, causing the person to writhe or twist into exaggerated positions. In late stage HD, chorea may subside. Some persons with HD experience little chorea and instead primarily experience slowing, stiffness, and poor coordination.

### Treating Chorea

In the early stages of HD, a person may be unaware of his or her chorea or they may incorporate the movements into their voluntary actions. If the chorea is mild, or if the person is not bothered by it, treatment can be limited to strategies such as balance training. However, if the person with HD is distressed by their chorea; if severe chorea is causing falls and accidents; or if it significantly interferes with quality of life, there are medications available which may reduce or control the involuntary movements.

Several types of medications can be used to control chorea. They will not slow the progress of HD and they have the potential to cause significant side effects. It is important to work closely with a physician or neurologist familiar with HD when considering chorea treatments. The types of drugs used to treat chorea will change over time and each person should consult with their primary care provider or a neurologist experienced with HD to determine what course of treatment/medications will work best for their individual condition.

### Impairment of voluntary movements

HD also affects voluntary movements and muscle control. Persons with HD often make movements that are exaggerated in size. In addition, they have trouble maintaining an ongoing movement. This may cause them to drop things or to dip at the knees when walking. Although chorea is the most obvious movement problem associated with HD, many persons with HD are much more disabled by these impairments in voluntary movements than by chorea. As the disease progresses movement slows and the person with HD loses coordination and small motor control. Walking becomes slower and more poorly coordinated and falls become more common. Diminished muscle control will also cause problems with swallowing, and speech will become slurred and harder to understand. In late stage HD a person will not be able to walk or to care for him or herself and their speech becomes impossible to understand even though they can understand much that is said to them.

## Treatment

Addressing the impairment of voluntary movements in HD requires a broad-based approach. While there are currently no treatments which can halt the advance of the disease, Physical Therapy (PT), Occupational Therapy (OT), Speech Therapy, Nutrition, and Assistive Devices may make it easier to adjust to the changing capabilities of the person with HD and prolong quality of life. For example, an OT may recommend putting corner guards on furniture or installing handrails in the home to help prevent bruises or falls while a Speech-Language Pathologist may introduce exercises to help maintain clarity of speech or swallowing techniques to aid in eating.

## Cognitive Disorders

HD is more than just a movement disorder. It also affects the ability of the brain to understand, organize and retain information. Changes in cognition (the ability to think) can be an early indicator of HD and will progressively affect cognitive functions such as organizing and prioritizing, controlling impulses, beginning and ending activities, creative thinking and problem solving. The person with HD may become forgetful, distracted or reckless.

Changes in cognition can be one of the hardest aspects of HD to accept. It can become a source of great frustration to both the person with HD and to their family. It can be hard to accept that disorganization and forgetfulness are symptoms of the disease and that the person with HD cannot

simply “try harder.” However, simple strategies and techniques exist which have improved the quality of life for many HD families, including:

- Maintaining familiar routines
- Breaking tasks into small steps
- Avoiding open-ended questions
- Adopting “to-do” lists and calendars
- Using patience and understanding

## Emotional/Behavioral Disorders

Among other things, HD causes progressive damage to the nerve cells in the brain that regulate thoughts and feelings. These unregulated emotions, caused by the disease, may cause mood swings and irritability. The person with HD may overreact to everyday events and may say cruel things or behave aggressively because of the changes in their brain. It is important to know when it is the disease “talking” and not the person.

### Treatment

Depression is also common in HD. It appears to be directly related to the brain disorder and it typically responds very well to standard treatments, though a person with HD may be very sensitive to the side effects of medication and should have his or her dosages carefully monitored by a doctor. The types of drugs used to treat depression will change over time and each person should consult with their primary care provider or a neurologist experienced with HD to determine an individual course of treatment.

## Being at-risk for HD

---

Each child of a person with HD has a 50/50 chance of inheriting the genetic variant that causes the disease. This knowledge affects people in different ways. Some choose not to think or talk about their status. Others think about it constantly. Some choose to live in the moment. Others try to live a balanced life, one day at a time. There is no one way to live at-risk for HD. Each individual must decide how to incorporate their at-risk status into major life choices, such as marriage, family planning and career decisions. Various resources are available for persons at-risk including social workers, genetic counselors, mental health professionals and neurologists who can help assess whether any symptoms or concerns might be due to HD or to other causes.

## Genetic Testing

---

Soon after the HD gene was identified in 1993, a direct gene test was developed which allows a person to find out if they carry the genetic variant and will someday develop the disease. While the test is very accurate, it cannot determine when the symptoms of the disease will begin or how severe the symptoms will be. A person without symptoms who tests positive for the gene may remain unaffected for many years.

Genetic testing presents persons at-risk for the disease with a difficult choice given the current absence of an effective treatment or cure. Many see no benefit in knowing that they will someday develop the disease. Others want an end to uncertainty so that they can make informed choices about the future. The decision whether or not to test is intensely personal and there is no “right” answer.

The Huntington’s Disease Society of America recommends that a person at-risk who is



considering genetic testing do so at a genetic testing center that follows HDSA's protocol for genetic testing. A complete list of these centers can be found on the HDSA national web site ([www.hdsa.org](http://www.hdsa.org)). Testing at these centers involves sessions with professionals who are knowledgeable about HD. To read more about the genetic testing process, please go to [www.hdsa.org/2016GTprotocol](http://www.hdsa.org/2016GTprotocol). The genetic test itself takes several weeks and uses a blood sample.

Genetic testing for children is typically prohibited before age 18, as the child may not understand the full implications of testing and may be vulnerable to pressure from others. However, a child under the age of 18 may be tested to confirm a diagnosis of Juvenile Onset HD after a thorough neurological exam.

Individuals or couples considering prenatal testing are advised to seek genetic counseling prior to a pregnancy. Many reproductive options are available to individuals affected by or at risk for HD, of which prenatal testing is one. Samples for prenatal analysis of the HD gene may be obtained in two ways: by chorionic villus sampling at 10-12 weeks of pregnancy, or by amniocentesis at 14-20 weeks.

Some couples may also desire preimplantation testing of a fertilized embryo. This requires the use of fertility drugs and other procedures available only at specialized *in vitro* fertilization centers. This procedure has a non-disclosing option as only those fertilized eggs that are gene negative are used. Couples planning a family should consult with a genetic counselor in order to explore the option that will be right for them.

# The Search for Effective Treatments and a Cure

---

The key to finding effective treatments and, ultimately, a cure for HD is research. HDSA remains one of the largest non-government funding sources of HD research in the United States and is the only organization dedicated to the care and cure of HD. But new treatments for HD cannot be approved without the participation of HD patients in clinical trials. Clinical trials are needed to define the safety, effectiveness and correct dose for new treatments. For more information go to [www.hdsa.org/research](http://www.hdsa.org/research). To find a clinical trial near you visit [www.HDtrialfinder.org](http://www.HDtrialfinder.org), an easy to use online matching service.

HDSA also supports a growing number of HDSA Centers of Excellence that serve as the cornerstone of HDSA's care program. An HDSA Center of Excellence is a regional multispecialty clinic that provides medical and social services to those affected by HD and their families. Services include but are not limited to medical care, genetic testing/counseling, medical and social services as well as access to clinical trials for potential treatments. To locate your closest HDSA Center of Excellence, please visit [www.hdsa.org](http://www.hdsa.org).

Together, HDSA's research and clinical care programs are contributing to speeding effective therapies to the HD community.





## Huntington's Disease Society of America

National Office  
505 Eighth Avenue, Suite 902  
New York, NY 10018  
Phone: 212-242-1968  
800-345-HDSA (4372)  
Fax: 212-239-3430  
Email: [hdsainfo@hdsa.org](mailto:hdsainfo@hdsa.org)  
[www.hdsa.org](http://www.hdsa.org)

