Huntington's Disease

A Family Guide

Huntington's Disease Society of America

The Huntington's Disease Society of America (HDSA) is dedicated to

- promoting and supporting research to find a cure for HD
- helping people and families affected by the disease, and
- educating the public and healthcare professionals about HD

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Family Guide Series

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Overview

Huntington's Disease (HD) is a hereditary brain disorder that affects people of all races all over 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 which are a common symptom of HD. In the United States, HD occurs in about 1 in 10,000 people. Currently about 30,000 people in the U.S. have HD and up to 200,000 are at risk.

In 1993, after a ten year search, scientists identified the gene that causes the disease. Since that breakthrough discovery, research has gained in momentum and much is now understood about HD and how it affects nerve cells in the brain. While researchers are actively looking for a treatment that can delay the onset or slow the progression of HD, medications are available that may help with some symptoms.

What is HD?

HD is a "neurodegenerative disease," which means that it causes the 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 also 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 forgetfulness, 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 Facets of HD" section beginning on page 8.

Symptoms appear gradually and people with HD can maintain their independence for 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 or your local HDSA chapter.

Inheritance

HD is a disease of families. Though everyone is born with the HD gene, the disease is caused by an abnormal copy of the gene that is passed from parent to child. It is not contagious in any way. Only a person who is born with the abnormal gene can ever get the illness or pass it on to their children. Every person who carries the abnormal copy of the gene will eventually develop symptoms, if they live long enough.

Every child born to an affected parent has a 50% chance of being born with the gene that causes the disease. Males and females have the same chance of inheriting the abnormal gene. Those who have not inherited the gene will not get the disease and they cannot pass it on to their children. HD does not "skip" generations.

Juvenile Huntington's Disease (JHD)

In approximately 10% of cases, HD affects children or adolescents. The symptoms of JHD are somewhat different than adult onset 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 previous had. JHD typically progresses more rapidly than adult onset HD. A variety of materials on JHD are available from the HDSA website, www.hdsa.org, or by calling 800-345-HDSA.

Clinical Diagnosis

Although the abnormal gene is present from birth, a clinical diagnosis of HD indicates that symptoms have started. A diagnosis of HD can only be accomplished by a comprehensive neurological evaluation that is best peformed by a specialist in HD or in neurological 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 HD gene, is not enough, by itself, to confirm a diagnosis of clinical HD.

For some, a diagnosis of HD 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 an HD support group, 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 thinking

through problems and often, a depressed or irritable mood. Medications are often effective in treating depression or other emotional problems. The effects of the disease may make the person less able to work at their customary level and less functional in their regular activities at home.

In the ***middle stage***, the movement disorder may become more of a problem. Medication for chorea may be considered to provide relief from involuntary movements. Occupational and physical therapists may be needed to help 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 do.

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.

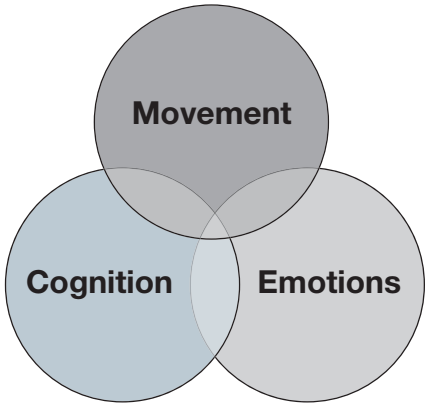
In all stages of HD, weight loss can be an important complication that can correspond with worsening symptoms and should be countered by adjusting the diet and maintaining appetite.

Living with HD

The Family

HD is a disease of individuals and also of families. A diagnosis of HD can draw families together but it also brings up many hard questions. How will the family care for the sick individual? Who else is at risk for the disease? Should they be tested? When should end-of-life decisions be made? How much should young children be told and when? There are no easy answers. Family and individual counseling or participation in HD support groups or HDSA chapters may help family members voice their feelings and concerns with people who share their experiences.

The Three Facets of HD



- Movement
- Cognition
- Emotions and Behavior

Understanding the wide-ranging impact of HD on an individual can help family and friends remain supportive of the person as he or she faces the challenges of the disease. Over time, HD affects all facets 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. Early signs of HD may include clumsiness, loss of balance and fidgeting. Problems with movements may include quick involuntary movements,

known as chorea, twisting postures known as dystonia, and reduced speed and accuracy of fine movements. The movement disorders of HD are often accentuated 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 individuals with HD experience little chorea and instead primarily experience slowing, stiffness, and poor coordination.

Treating chorea

Chorea is probably the most common symptom of HD. In the early stages, people with HD may be unaware of their 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. These medicines do not slow down 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. HD patients 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 patients are much more disabled by these impairments in voluntary movements than by the chorea. As the disease progresses movement slows and the patient

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 the speech of a person with HD will become slurred and harder to understand. Late stage HD patients are not able to walk or to care for themselves 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 changing capabilities 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 causes more than movement disorders. It also affects the ability of the brain to

understand, organize and retain information. Changes in cognition (the ability to think) can be an initial indicator of HD. HD progressively affects 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 patient 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 patient may overreact to everyday events. People with HD may say cruel things or behave aggressively because of the changes in their brains. It is important to know when it is the disease "talking" and not the patient.

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 HD patients may be very sensitive to the side effects of medication and should have their 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 a course of treatment for their individual condition.

Being at-risk for HD

Each child of a person with HD has a 50/50 chance of being born with the mutated gene that causes the disease. This knowledge affects people in different ways. Some choose not to think or talk about their at risk 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 people at risk for the disease, including social workers and support groups, genetic counselors, and neurologists who can help assess whether any symptoms of concern 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 abnormal HD gene 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. People without symptoms of HD who test positive for the gene may remain unaffected for many years.

Genetic testing for HD presents people at risk for the disease with a difficult choice, given the current absence of an effective treatment or cure. Many people 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 to test or not is intensely personal and there is no "right" answer.

The Huntington's Disease Society of America recommends that at risk individuals who are considering genetic testing do so at a genetic testing center that follows HDSA guidelines for genetic testing. A complete list of these centers can be found on the HDSA national web site (www.hdsa.org). Testing procedures at these centers involves sessions with professionals who are knowledgeable about HD and generally include one session devoted to each of the following: genetic counseling, a neurological exam, a psychological interview, discussion of the results and follow up. The genetic test

itself takes several weeks and is performed on 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.

For couples planning a family, prenatal testing of an at-risk fetus is an option. This can be done through a direct genetic test or through a "nondisclosing" variation of the test that approximates the risk that the fetus is carrying the HD gene without disclosing the genetic status of the at-risk parent. A second option is PGD or pre-genetic diagnostic testing in which eggs that have been fertilized are tested for the abnormal HD gene within a few days of fertilization and only those without the mutated gene are re-implanted in the mother. This procedure is also a non-disclosing option as only those blastocysts that are gene negative are used. Other options include amniocentesis, to identify fetuses that carry the mutated gene for possible termination, and adoption. Couples planning a family should consult with a genetic counselor in order to explore the option that may 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. In addition to supporting the prestigious HDSA Coalition for the Cure, HDSA has also created and funds a pipeline for drug discovery that includes basic research, drug discovery and development, and clinical trials that can lead to FDA approved drugs for treating HD. Advances in research are rapid. Please visit the HDSA national web site at www.hdsa.org to read about the research being supported by HDSA today.

HDSA also supports 21 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, social services and access to clinical trials for potential treatments. To locate your closest HDSA Center of Excellence, please visit our web site at www.hdsa.org.

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

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

HDSA is a national not-for-profit organization founded to help individuals with Huntington's Disease and their families. HDSA is often the first place that people go for information and assistance when coping with the effects of HD. HDSA publishes and distributes a wide variety of informational materials covering care, treatment, research and related topics.

There are more than 38 HDSA Chapters and Affiliates throughout the country. They help people new to the HD community to connect with others who share their concerns. HDSA Chapters also provide information about local resources, knowledgeable physicians and other health professionals, genetic testing centers, support groups and long-term care facilities. In most chapters, a social worker is available for information and support.

HDSA also offers more than 150 support groups across the nation. To find the nearest HDSA chapter, affiliate, Center of Excellence, genetic testing center or support group, please visit the HDSA national web site at www.hdsa.org.

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