

# Huntington's Disease

## A Guide for Families

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## I. Introduction

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 physician who published a description of what he called "hereditary chorea" in 1872. From the Greek word for "dance," chorea refers to the involuntary movements which are among the common symptoms of HD.

Until quite recently, little was known or published about Huntington's Disease. Yet in the last 20 years, much has been learned about the causes and effects of HD and about treatments, therapies and techniques for managing the symptoms of the disease. In 1993, after a ten-year search, scientists found the gene that causes HD, and important advances have flowed from this dramatic discovery. Many scientists are actively engaged in the search for effective treatments to stop or reverse the effects of HD, and eventually to cure it altogether.

HD is a degenerative disease whose symptoms are caused by the loss of cells in a part of the brain called the basal ganglia. This damage to cells affects cognitive ability (thinking, judgment, memory), movement and emotional control. Symptoms appear gradually, usually in mid-life, between the ages of 30 and 50. However, the disease can strike young children (see Juvenile HD, section IV.) and the elderly.

In most cases, people can maintain their independence for several years after the first symptoms of HD appear. A knowledgeable physician can prescribe treatment to minimize the impact of symptoms. Allied health professionals, such as social workers, occupational and physical therapists, speech-language pathologists (speech therapists), and nutritionists, can all play a useful role in maximizing abilities and prolong independence.

## II. Inheritance

HD is a family disease for two reasons. First, it is passed from one generation to the next by the transmission from the parent to a child of a "mutated" (altered) gene (see VIII). Each child of an affected parent has a one in two, or 50 percent, chance of inheriting the gene that causes HD, and is said to be "at risk." People who carry the gene will eventually develop Huntington's unless they die of some other cause before the onset of symptoms.

HD occurs in approximately 1 in 10,000 people in the United States. Currently, about 30,000 people in the U.S. have HD and a further 150,000 are at risk.

Males and females have an equal chance of inheriting the gene from an affected parent. Those who do not inherit the gene will not develop the disease, nor will their children; HD does not "skip a generation." Genetic testing is available to determine whether or not a person carries the gene for HD (see VII).

### III. The Family

HD is also a family disease because of its impact on every family member. As the disease progresses, the family role of the affected person will gradually change. The spouse or partner will have to assume more and more of the house-keeping, decision-making and parenting duties which his/her partner may no longer be able to fulfill. In addition, the spouse or partner will most likely become the primary care giver.

Children and adolescents must face living with a mother or father who is ill and whose behavior may be erratic. They may even be asked to participate in the parent's care. For those parents, telling children about HD can pose difficult questions. Should a child/children be told about HD? If so, at what age? When is a child old enough to cope with the idea of being at risk for HD?

There are no easy answers, particularly since children develop at different rates and each family situation is different. Generally, it is a good idea to be as open as possible without being alarmist, and to convey the facts bit by bit. This way, a child can develop a gradual awareness of HD rather than being suddenly overwhelmed by information.

### IV. Symptoms and Stages of HD

*It is not helpful to treat HD as a shameful family secret, as a child or adolescent will find out about it eventually. Withholding the truth can lead to mistrust and resentment.*

The symptoms of HD vary widely from person to person, even within the same family. For some, involuntary movements may be prominent even in the early stages. For others, these may be less evident and emotional and behavioral symptoms may be more obvious. The following are common features of HD:

#### Emotional/Behavioral Symptoms

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Depression, irritability, anxiety and apathy are often encountered in HD. Some people can experience depression for a period of months or even years before it is recognized to an early symptom of Huntington's. Behavioral changes may include aggressive outbursts, impulsiveness, mood swings, and social withdrawal. Often, existing personality traits will be exacerbated by HD, e.g., a person who had a tendency to irritable. Schizophrenia and other serious psychiatric problems are common in HD but do occur.

#### Cognitive/Intellectual Symptoms

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Slight intellectual changes are often the first signs of cognitive disturbance. These may involve reduced ability to organize routine matters or to cope with new situations. Short-term memory loss may occur while long-term memory generally stays intact. Work tasks become more difficult.

## Motor Symptoms

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Physical symptoms may initially consist of "nervous" activity, fidgeting, twitching, or excessive restlessness. Handwriting may change and facial grimaces may appear. Day-to-day skills involving coordination and concentration, such as driving, become more difficult.

These initial symptoms will gradually develop into more marked involuntary movements of the head, trunk and limbs - which often lead to problems with walking and balance. Speech and swallowing can become impaired. Movements generally tend to increase during voluntary effort, stress or excitement, and decrease during rest and sleep.

## The Stages of HD

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Though the pattern and severity of symptoms vary from person to person, the course of HD can be roughly divided into three stages.

Early in the disease, manifestations include subtle changes in coordination, perhaps some involuntary movements, difficulty thinking through problems, and often, a depressed or irritable mood. At this stage, medications are often effective in treating depression and other emotional symptoms. It is a good time to begin planning for the future. Financial plans should be made and legal documents (a Living Will, for example) drawn up. HDSA chapter social workers and family service coordinators can help to determine what is needed.

In the middle stage, involuntary movements (chorea) may become more pronounced. A staggering gait can sometimes be mistaken for drunkenness (it can be helpful to carry documentation that clearly refers to a diagnosis of Huntington's Disease). Speech and swallowing will begin to be affected. It is important to consult a speech language pathologist (speech therapist) who will be able to offer suggestions and strategies for improving communication and swallowing abilities. Likewise, occupational and physical therapists can develop programs to help maintain the highest level of functioning and thereby improve quality of life.

Thinking and reasoning skills will also gradually diminish. At this stage it may become increasingly difficult to hold a job and to carry out household responsibilities. Here again, simple strategies may be employed to help decrease frustration, increase functioning, and prolong independence. For example, disorientation and short-term memory loss can be addressed by labeling drawers, maintaining a daily routine and posting a calendar listing appointments and events.

People with Late-stage HD may have severe chorea, but more often have become rigid. Choking on food often becomes a major concern, as does weight loss. At this stage people with HD are totally dependent on others for all aspects of care, can no longer walk, and are not able to speak.

Although cognitive abilities are severely impaired, it is important to remember that the person is generally still aware of his/her environment, remains able to comprehend language, and retains an awareness of loved ones and others. He/she may continue to enjoy looking at photographs and hearing stories of family and friends.

People do not die from HD itself but rather from complication of the disease, such as choking or infection. Death generally occurs about 15 to 20 years after onset.

## V. Diagnosis

A clinical diagnosis of HD can only be accomplished by a comprehensive examination which generally entails a series of neurological and psychological exams and a detailed family history. MRI (magnetic resonance imaging) or CT (computerized tomography) scans may be included in the exam but the findings from these procedures are not sufficient to form a diagnosis.

Similarly, a genetic test may be used to help confirm, or rule out, a diagnosis of HD. However, a positive test result (indicating the presence of the HD gene) is not sufficient in and of itself (i.e., without a neurological exam) to confirm a diagnosis of HD.

It is best to see a neurologist who is very familiar with HD, as the symptoms can mimic those of other disorders such as Parkinson's disease or alcoholism. Referrals to knowledgeable professionals can be provided by your local HDSA chapter.

For some, a diagnosis of HD can be a relief. It may provide an explanation for why their memory is not quite as sharp as it used to be or why they are feeling irritable or depressed. Others find the news very upsetting.

It is not uncommon for people to be in a state of "denial" when they are first diagnosed with HD. Regardless of their initial reaction, it can help to discuss the situation with others, either in a support group, with an HDSA social worker, or with another counselor or therapist.

## VI. Juvenile Huntington's Disease

In approximately ten percent of cases, Huntington's Disease affects children or adolescents. Children most often inherit the disease from their fathers (adult-onset HD is inherited from both parents with the frequency). The symptoms of the juvenile form, or Westphal variant, of HD are somewhat different from adult-onset HD.

Initial symptoms usually involve slow, stiff and awkward walking and talking, choking, clumsiness and falling. Later, the child may become slow to respond and performance at school may become erratic. The course of the juvenile variant is generally more rapid than adult-onset HD. The booklet, *Living with Juvenile Huntington's Disease* and the videotape, *Claudia's Challenge*, both provide more information on juvenile HD and are available from HDSA.

## VII. Being At Risk For HD

Being at risk for Huntington's Disease affects different people in different ways. Some choose not to think or talk about their at risk-status, even to the point of shunning other family members. Others think constantly about being at risk and about the possibility of developing HD. This can have an insidious influence and may lead to behavior which is impulsive or self-destructive. Still others are able to find a balanced approach to their at-risk status and approach decision-making in this way.

Being at risk for HD influences major life choices such as marriage, family planning and career decisions. It can also have a pervasive influences on everyday activities. An episode of clumsiness, twitching or forgetfulness , such as everyone experiences form time to time, may be seen as a potential symptom of HD and take on nerve-racking significance.

Many people come to accept the uncertainty of being at risk for HD, especially in the absence of an effective treatment or cure for the disease. Indeed, faced with the choice, most prefer to live with this uncertainty rather than taking a test which could remove hope by confirming that they will develop HD.

For others, genetic testing for HD offers a chance to end the uncertainty and to gain information which they believe will enable them to make choices about the future.

## VII. Genetic Testing

Soon after the Huntington's Disease gene was found in 1993, a test was developed which enabled people to find out if they were carrying the gene that causes HD. Earlier tests were based on a process of "linkage analysis" which required blood samples from several family members. The new "direct gene test" is much more accurate and requires blood only from the individual taking the test.

The HD gene was found to contain a specific section that was expanded in people with HD. In all people, this stretch of genetic material, or DNA, contains a pattern of so-called "trinucleotide repeats." Nucleotides are the building blocks of DNA and are represented by the letters C, A, G, and T. In most people, the repeated pattern, CAG, occurs 30 times or less. In Huntington's Disease, it occurs more than 40 times. By analyzing a person's

DNA and counting the number of CAGS, it is possible to tell if that person will develop HD. The test cannot predict age of onset.

The decision to undergo genetic testing is an intensely personal one and one that cannot be taken lightly. Everyone has their own circumstances to take into consideration, and there are no "right" or "wrong" answers. Testing should never be forced upon an at-risk individual. Children are generally not able to consider the full implications of testing and may be vulnerable to pressure from others. Therefore, the minimum age requirement is usually 18.

Various resources are available to assist you in making this decision. HDSA chapter social workers and genetic counselors at testing centers can help, and there may be a support group for people at risk for HD in your area.

The Huntington's Disease Society of America recommends that at-risk persons who wish to undergo presymptomatic testing do so at an HD testing center. The testing centers involve teams of professionals who are knowledgeable about HD, and a list of these centers is available from HDSA.

The testing procedure involves sessions with various professionals. It typically includes one session devoted to each of the following: genetic counseling; a neurologic exam; a psychological interview; discussion of the results; and follow-up. The genetic test itself is a blood test.

The purpose of the preliminary sessions is to ensure that the person understands the potential implications of his/her genetic status and is prepared to receive the results. The neurologic exam will determine if any early symptoms of HD are present. If the person is found to be symptomatic, he/she will be offered the option of discontinuing the testing procedure.

It is important to note that presymptomatic testing for HD cannot determine when the disease will begin or its course or severity. People who test positive for the gene may remain healthy for many years. HD can only be diagnosed by a neurological exam.

For couples planning a family, prenatal testing of a fetus is also an option. A "nondisclosing" variation of the prenatal test can also be done using linkage analysis. Instead of examining the gene, this method of testing compares patterns of chromosomal inheritance in several family members with the chromosomes inherited by the fetus. In this way, the approximate risk that the fetus is carrying the HD gene can be determined without disclosing the genetic status of the at-risk parent.

## **IX. Treatment**

Treatment for HD takes many forms. While current treatments do not alter the course of HD, medications can be effective in treating common symptoms such as depression and anxiety, for example. Involuntary movements can also be lessened by medication. Some drugs have significant side effects, however, so it is important that these be explained by the physician before the treatment begins.

Some doctors may prescribe drug treatment when it is not absolutely necessary. In many cases, people with HD do better when medication is kept to a minimum. Often, drugs that are effective at one stage of the disease may not be effective at another.

For these reasons, it is preferable to locate a neurologist with expertise in HD. Not all neurologists are familiar with the disease. Though a family physician is not likely to have much experience with HD, that physician should remain involved in ongoing care and treatment. The person with HD and family members play a critical role in monitoring and assessing the effectiveness of any care and treatment.

*A Physician's Guide to the Management of Huntington's Disease* contains useful and detailed information on the treatment of specific symptoms of HD and is available from HDSA.

It is also helpful to consult a physical or occupational therapist and a speech-language pathologist (speech therapist) so that strategies that can have a positive and lasting impact on the quality of life can be implemented early.

Nutrition is important in everyone's life, but takes on added significance in HD. People with HD require an unusually high number of calories to maintain their body weight. Maintaining, or even gaining, weight can help reduce involuntary movements and other symptoms, particularly in the later stages of HD. Nutritional supplements such as Ensure can help, and a nutritionist can offer other valuable suggestions.

Often the best advice and emotional support one gets is from someone who "has been there." The mutual support given and the knowledge shared are the reasons that many find HD support groups to be an important part of their lives. Support groups are located in most states and HDSA can help you locate the one closest to you.

## **X. The Search For A Cure**

The key to better treatments and an eventual cure for HD is research. There have been several exciting breakthroughs in recent years, notably the HD gene discovery of 1993. Since then, certain brain proteins have been discovered which appear to interact with huntingtin, the protein expressed by the HD gene. Research is under way to determine how these substances combine to cause the symptoms of HD, and to find ways of stopping this interaction as a possible means of treatment.

After the gene discovery, an international coalition of scientists, known as the Huntington Study Group (HSG), was formed to conduct basic and clinical research in a coordinated fashion. HSG sites combine research facilities with teams of doctors with expertise in treating HD. The group has begun to test new drugs which could potentially lead to effective treatments for Huntington's Disease.

In 1997, the Huntington's Disease Society of America established the HDSA Coalition for the Cure, a consortium of 14 top laboratories in North America and Europe. Coalition investigators focus on four key areas of study: animal models, cell models, biochemistry

and cell biology. Through HDSA funding, semi-annual meetings and the sharing of data and ideas, the Coalition is accelerating the pace of HD research.

## **XI. Huntington's Disease Society of America**

With over 30 chapters throughout the country, the Huntington's Disease Society of America is often the first place people go for information or assistance in coping with the effects of HD. HDSA chapters can provide information about local resources, including knowledgeable physicians and other health professionals, genetic testing centers, support groups, and long-term care facilities. In most cases, a chapter social worker is available for information and support.

HDSA publishes and distributes a wide variety of informational materials, including newsletters, books, booklets, brochures and videotapes covering care, treatment, research, and related topics.

The Society's commitment to research is demonstrated by its continued increases in funding to HDSA research programs each year. This year the Society will dedicate over \$2.5 million for medical and research initiatives including the HDSA Coalition for the Cure, the HDSA Grants and Fellowships program, and the HDSA Centers of Excellence for Family Services and Research. For more information about how you can help or to find out more about HDSA, please write to:

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