

National Society of Genetic Counselors Fact Sheet

Huntington's Disease and Predictive Genetic Testing

What is Huntington's disease?

Huntington's disease is an inherited brain disorder that is caused when specific cells in the brain die. Symptoms include **mood and behavior disturbances**, including depression, apathy, irritability, anxiety, and obsessions; **cognitive decline**, such as loss of memory and inability to focus, plan, recall or make decisions; and **physical deterioration** due to large involuntary movements, which cause loss of coordination and difficulty walking, talking, and swallowing. Symptoms typically start in middle age between 30-50 years, although they can also develop in younger and older people and become worse as the disease progresses until death occurs. While the physical symptoms of Huntington's disease become incapacitating, the emotional and cognitive features can be equally debilitating, often causing loss of job, relationships, and independency in life.

Currently, there is no cure for Huntington disease, although medications are available to help manage symptoms. The effectiveness of these drugs varies from patient to patient and they tend to become less effective over the course of the illness.

How common is Huntington disease?

Huntington's disease affects one in every 10,000 persons or approximately 30,000 people in the United States. While it is not a common disease, its impact is far-reaching. It's estimated that for every person who has Huntington's disease, the disorder impacts another 20 individuals including friends, caregivers, and family members who may be at risk for developing the disease themselves.

What causes Huntington's disease?

HD is caused by a mutation in a gene that all humans have. A mutation is a change in a gene that causes a disease. Everyone has two copies of the Huntington's disease gene but only those who have a **genetic mutation** in one copy of the gene may develop the disease.

The mutation involves an increase in a small segment of DNA, called a **CAG repeat**. Normally,

individuals have approximately 17 CAG repeats but people who have 36 CAG repeats or more are considered **mutation-positive** and will develop Huntington disease in their lifetime. Although, individuals with 36-39 CAG repeats often have a later age of onset than what is typical observed. Individuals with these so-called **reduced penetrance alleles** have elderly onset, if symptoms occur at all. Persons with 26 CAG repeats or fewer are **mutation-negative** and will never develop the disease. Some individuals fall in the middle and have what is called an **intermediate allele** (27 to 35 CAG repeats), meaning they will usually not develop the disease but there may be a chance their children will.

A “Family Disease”

Huntington’s disease is often described as a family disease. It is a genetic disorder that is inherited in an **autosomal dominant** manner. Children whose parents are mutation-positive have a 50 percent chance of developing the disease when they are adults. They will either inherit the gene with the mutation or they will get the normal gene from their parent. Children of parents who are mutation-negative are not at risk. Individuals who are at-risk of the disease can undergo **predictive genetic testing**.

What is predictive genetic testing?

Predictive genetic testing involves a simple blood test to detect whether or not the individual has the genetic mutation that causes the disease. Huntington’s disease was the first disease for which predictive testing was offered. People over 18 years old who have a family member affected with Huntington’s disease but do not yet have symptoms of the disease are eligible for testing. A neurological exam is also performed to look for early symptoms. Anyone considering predictive testing should meet with a **genetic counselor**.

The role of genetic counseling

The role of genetic counseling is to provide patients and families with the information and support they need in order to make an **informed decision** about predictive testing for Huntington’s disease. The decision whether to have predictive testing is extremely personal and it’s important to take your time and make an informed choice. A genetic counselor can help you talk through all the factors to

consider.

During the genetic counseling sessions you'll discuss many issues, including the potential benefits and harms of testing, your motivations for testing, your expectations about the test result, the impact either a mutation-positive or negative result might have on your life, and strategies for dealing with the test result. A genetic counselor will also help you identify other issues, such as the impact testing may have on your relationship with your spouse, children and extended family members, your career, and your insurance and finances. For example, results from predictive testing could make it difficult to purchase life, disability or long-term care insurance.

The pros and cons of predictive testing

Since predictive testing became available, many people have struggled with the question - *should I have predictive testing or not?* This decision is extremely complex, with many factors weighing in as pros and cons. People in favor of testing often cite their motivations to be an increased ability to plan for the future and make informed reproductive decisions, the relief of uncertainty and worry, the desire to learn their children's risk status, the ability to participate in research and clinical trials, and simply for the sake of wanting to know. Reason given for why some people prefer not to have the test include concern over the possibility of adverse emotional reactions, fear of receiving a mutation-positive result, concerns over genetic discrimination, the lack of a cure, the preference of living with hope that they will not develop the disease and merely the desire not to know.

Often people who have not had the experience of living in a family affected by Huntington's disease underestimate the impact of predictive testing and assume that all persons who are at-risk will have the test. Although there are many ways in which this genetic information can be useful, it often comes down to whether or not someone feels like they can live with the test result they receive. The decision to know your mutation status is extremely difficult when you have seen generations of your family affected by the disease and have helped care for affected loved ones. The complexity of this decision is likely why only 10% of people living at-risk in the USA decide to pursue predictive testing.

The emotional impact of testing

The emotional impact of receiving predictive test results can be difficult to anticipate and some individuals may experience negative reactions, although most people experience a positive impact on their psychological wellbeing.

Individuals who receive a mutation-positive result may initially experience feelings of depression, anger, fear or despair. Many people struggle with not knowing when the first symptoms will appear, and may start to “symptom watch” and wonder if occasional clumsiness or forgetfulness are early signs of the disease. With time and support, most people come to accept their result.

Individuals who receive a mutation-negative result may initially experience feelings of joy and relief. But some struggle with “survivor’s guilt” because they escaped the disease while other family members did not. They may also feel an increased responsibility to care for affected family members. It’s important to acknowledge that individuals who receive a negative result may also benefit from additional support following testing.

How can I get more information?

If you would like more information on Huntington’s disease, please visit the Huntington’s disease Society of America’s website at <http://www.hdsa.org>. If you are interested in learning more about predictive testing, find a genetic counselor in your area by using the National Society of Genetic Counselors’ “Find a Genetic Counselor” tool at www.nsgc.org/findageneticcounselor.

