Huntington’s Disease

A topic in the Alzheimer’s Association series on understanding dementia.

About Dementia

Dementia is a condition in which a person has significant difficulty with daily functioning because of problems with thinking and memory. Dementia is not a single disease. It’s an overall term — like “heart disease” — that covers a wide range of specific medical conditions, including Alzheimer’s disease. Disorders grouped under the general term “dementia” are caused by abnormal brain changes. These changes trigger a decline in thinking skills, also known as cognitive abilities, severe enough to impair daily life and independent function. They also affect behavior, feelings and relationships.

Brain changes that cause dementia may be temporary, but they are most often permanent and worsen over time, leading to increasing disability and a shortened lifespan. Survival can vary widely, depending on such factors as age at diagnosis and coexisting health conditions.

Huntington’s Disease

Huntington’s disease is a progressive brain disorder caused by a single defective gene on chromosome 4 — one of the 23 human chromosomes that carry a person’s entire genetic code. This defect is “dominant,” meaning that anyone who inherits it from a parent with Huntington’s will eventually develop the disease. The disorder is named for George Huntington, the physician who first described it in the late 1800s.

The defective gene codes the blueprint for a protein called huntingtin. This protein’s normal function isn’t yet known, but it’s called “huntingtin” because scientists identified its defective form as the cause of Huntington’s disease. Defective huntingtin protein leads to brain changes that cause abnormal involuntary movements, a severe decline in thinking and reasoning skills, and irritability, depression and other mood changes.

Prevalence

About 30,000 Americans — one in every 10,000 — have Huntington’s disease. An additional 150,000 to 200,000 are known to be at risk because they have a parent with Huntington’s.

Symptoms

The hallmark symptom of Huntington’s disease is uncontrolled movement of the arms, legs, head, face and upper body. Huntington’s disease also causes a decline in thinking and reasoning skills, including memory, concentration, judgment and ability to plan and organize.

In addition, Huntington’s disease brain changes lead to alterations in mood, especially depression, anxiety and uncharacteristic anger and irritability. Another common symptom is
obcessive-compulsive behavior, leading a person to repeat the same question or activity over and over.

**Diagnosis**

Scientists identified the defective gene that causes Huntington’s disease in 1993. A diagnostic genetic test is now available. The test can confirm that the defective gene for huntingtin protein is the cause of symptoms in people with suspected Huntington’s disease and can detect the defective gene in people who don’t yet have symptoms but are at risk because a parent has Huntington’s.

**Causes and Risk Factors**

The defective gene identified in 1993 causes virtually all Huntington’s disease. This gene codes a protein that scientists called “huntingtin” after linking it to Huntington’s disease. The huntingtin protein gene and all other human genes carry their biological blueprints in repetitions of simple chemical codes.

The huntingtin gene defect involves extra repeats of one specific chemical code in one small section of chromosome 4. The normal huntingtin gene includes 17 to 20 repetitions of this code among its total of more than 3,100 codes. The defect that causes Huntington’s disease includes 40 or more repeats. Genetic tests for Huntington’s disease measure the number of repeats present in an individual’s huntingtin protein gene.

Scientists don’t yet understand the normal function of huntingtin protein or how a few dozen extra repeats in its genetic blueprint lead to the devastating symptoms of Huntington’s disease. Researchers are eager to solve these mysteries not only to find the answer to Huntington’s but also because the solutions may offer important insights into a wide range of other brain disorders, including Alzheimer’s, Parkinson’s disease and amyotrophic lateral sclerosis (ALS).

**Outcomes**

Huntington’s is a progressive disease with symptoms and brain changes that gradually get worse. Symptoms usually develop between ages 30 and 50, but they can appear as early as age 2 or as late as 80. People with Huntington’s survive an average of 15 to 25 years.

**Treatment**

There is currently no cure for Huntington’s disease and no way to slow or stop the brain changes it causes. Treatments focus on managing symptoms. In 2011, a group of more than 50 international experts recommended the following treatments as first-line strategies for three of the disorder’s most troubling symptoms:

- **Chorea (involuntary movements):** Some experts begin treatment with an atypical antipsychotic drug. Others start with another type of drug recently approved by the U.S. Food and Drug Administration (FDA) specifically for Huntington’s.
- **Irritability:** For severe anger and threatening behavior, experts agree that an atypical antipsychotic drug is the best first-line approach. For less severe, nontreatment
irritability, experts recommend first trying a selective serotonin reuptake inhibitor (SSRI), a type of antidepressant.

- **Obsessive-compulsive thoughts and actions**: Experts also recommended SSRIs as the front-line treatment for these symptoms.

Other Huntington’s symptoms, such as anxiety, depression and insomnia, should also be treated according to generally accepted guidelines. Experts encourage people with Huntington’s to keep all their medical appointments and not to get discouraged if it takes their health team some time to find the best drugs and the most effective doses for them.

**Learn More**

For more information on Huntington’s disease and other topics in the Alzheimer’s Association series on understanding dementia, visit www.alz.org, or call our toll-free, 24/7 Helpline at 800.272.3900.

The Alzheimer’s Association is the world’s leading voluntary health organization in Alzheimer’s care, support and research.