BRAIN SPECIAL

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Highlighting Huntington disease

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Get the facts about one of the most common hereditary brain disorders.

By Amanda Perkins, MSN, RN

Huntington disease is an incurable, progressive, genetic disorder leading to the breakdown of nerve cells within the brain. Affecting men and women equally, it results in loss of muscle control, memory, and cognition, and is fatal for all patients diagnosed, often within 15 years of onset. Huntington disease is rare, with 30,000 Americans currently diagnosed and an additional 200,000 or more at risk for inheriting the disease, according to the Huntington's Disease Society of America (HDSA).

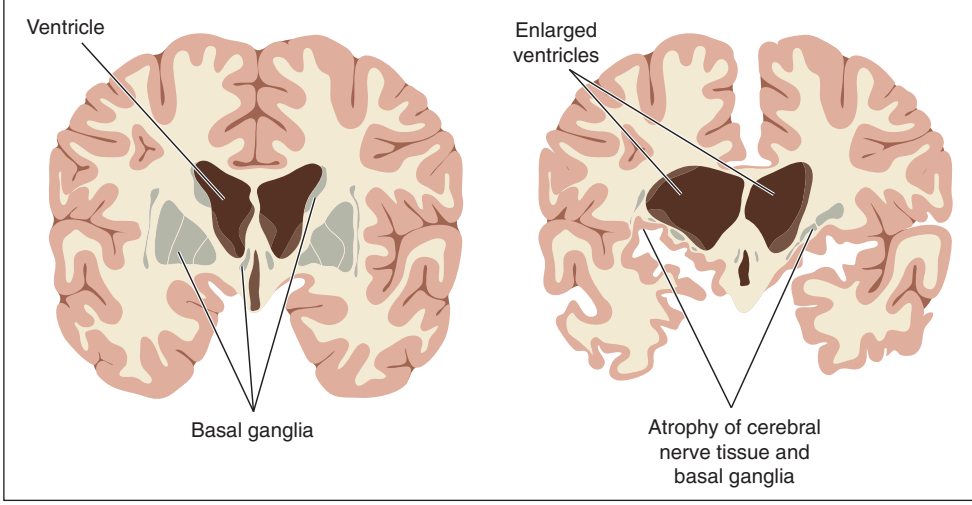
This article discusses anatomy and physiology, genetics, signs and symptoms, diagnosis, complications, and the management of Huntington disease.

Anatomy and physiology

Huntington disease affects the whole brain; however, some areas, such as the basal ganglia, striatum, and cerebral cortex, are affected to a greater degree (see *Brain atrophy in Huntington disease*).

The basal ganglia is a group of nuclei responsible for movement, language,

Brain atrophy in Huntington disease



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thought, emotion, and motivation, whereas the cerebral cortex is responsible for thought, perception, language, and memory. The basal ganglia is crucial for the performance of smooth, controlled movement. The striatum is a key component of the basal ganglia.

The cerebral cortex is the area of the brain in which the highest level of neural processing takes place. At the time of death, the brain affected by Huntington disease will have an average volume loss of 20%, mostly in the striatum and cerebral cortex; up to 70% of the nerve cells in some of the layers may be lost.

did you know?

The famous American folk musician and activist Woody Guthrie, who wrote the song "This Land is Your Land," was diagnosed with Huntington disease in 1952. After his death in 1967, his wife, Marjorie, founded what became the HDSA at a time when little was known about the disorder. She convinced President Jimmy Carter to create a commission to study the disease, which resulted in a cornerstone 1977 report. Read more at http://hdsa.org/about-hdsa/history.

Genetics

Huntington disease is caused by an autosomal dominant gene, meaning that each child of a parent with the disease has a 50% chance of carrying the gene and, ultimately, developing the disease. Basically, if a person has one copy of the defective gene, Huntington disease will develop. If a child doesn't inherit the defective gene, he or she won't develop Huntington disease and can't pass it on to his or her own children. It's interesting to note that approximately 1% to 3% of individuals diagnosed with Huntington disease don't have a family history of the disease.

This gene mutation, found on chromosome 4, is an expanded triplet repeat that codes for an abnormal version of the huntingtin protein, which accumulates in nerve cells and leads to their destruction. The exact function of the huntingtin protein is unknown, but research shows that it's widely distributed in the brain and associated with transcription, protein turnover, and energy production. Basically, this gene mutation results in the repetition of the base pairs cytosine (C),

adenine (A), and guanine (G) in the patient's DNA.

The number of repetitions can vary, with as little as 38 or as high as 121 repetitions. The number of CAG repetitions is relational to the onset of the disease, with increased numbers of repetitions linked to an earlier onset of symptoms. People who don't have Huntington disease can also have these repeats, but the number of repeats will be less than 35.

Signs and symptoms

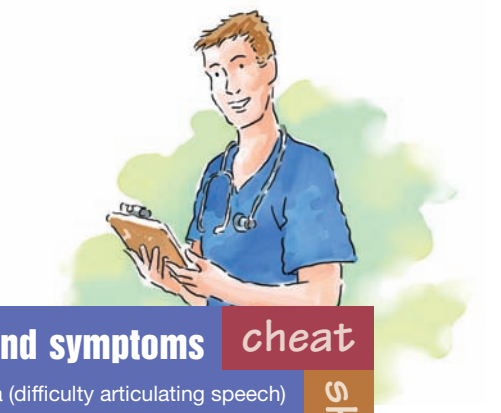
Patients with Huntington disease experience psychiatric, medical, and social problems. Signs and symptoms typically develop between ages 30 and 50. After symptoms develop, they progressively worsen over a span of 10 to 25 years. The rate at which signs and symptoms progress and the time at which they present varies. Generally, the cognitive signs associated with Huntington disease become apparent before movement problems.

The signs and symptoms that may be observed in Huntington disease include the following:

- dysarthria (difficulty articulating speech)
- abnormal eye movements
- stronger deep tendon reflexes
- anxiety
- obsessive compulsive behavior
- paranoia
- personality changes
- mood swings
- depression
- forgetfulness
- impaired judgment
- unsteady gait
- involuntary movements (chorea)
- slurred speech
- difficulty swallowing
- significant weight loss.

One of the earliest and most characteristic signs and symptoms of Huntington disease is involuntary, jerking movements of the limbs, torso, and face called chorea. In addition to chorea, some patients report that early signs and symptoms include mood swings, depression, irritability, difficulty driving, and problems with memory and decision making.

Individuals may be diagnosed with a mental disorder if they're unaware of a family history of Huntington disease.



Signs and symptoms cheat sheet

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Stages

Huntington disease can be divided into an early, middle, and late stage.

In the early stage, subtle changes in coordination may be observed. Additionally, the patient may start to develop chorea and depression, and have difficulty problem solving and an irritable mood. While in this stage, the patient may start to have difficulties completing tasks at work and home.

In the middle stage, patients may have more pronounced movement difficulties. The changes seen in thinking, reasoning, and speech continue to worsen in this stage and ordinary activities become increasingly difficult.

In the late stage, the patient will be completely dependent on others for care. Swallowing becomes very difficult and

choking is a major concern. As is the case with all patients who have difficulty swallowing, there's a risk of aspiration and pneumonia, which can develop as the result of aspiration. In this stage, one of two things can happen with chorea: it can become increasingly severe or it can diminish. These patients aren't able to walk or speak, but will still be able to comprehend language and retain awareness of friends and family.

Diagnosis and testing

The diagnosis of Huntington disease is made based on a history and physical, brain scans, and genetic testing. Neurologic assessments and lab tests may also be completed. In 1993, the gene that distinguishes Huntington disease was identified, making diagnosis easier and more effective. Genetic testing is done on a blood sample to look at an individual's DNA for the huntingtin gene and then measure the number of gene repeats.

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Genetic testing can be performed for individuals who are at risk for the development of Huntington disease. For example, the child of a parent with Huntington disease may choose to be tested before starting a family. Prenatal testing may also be performed.

Genetic testing for children under age 18 is prohibited because children can be pressured to take the test and may not fully understand the test and/or results. The only allowance to this rule is if a child is believed to have juvenile-onset Huntington disease (see *Juvenile Huntington disease*). Before genetic testing, the child will undergo a neurologic exam.

Genetic testing can be completed predictively—to assess a person's risk of developing Huntington disease—or diagnostically—to confirm a diagnosis. Before either type of testing, the individual considering genetic testing should meet with a genetic counselor. Counseling should be ongoing throughout the entire process, from pretesting through posttesting. Individuals undergoing genetic testing should understand how it works, the risks and benefits, and the consequences of test results. Additionally, these individuals should be educated about their ability to withdraw from testing at any time during the process. Informed consent is necessary before genetic testing.

The steps of the genetic testing process are pretest genetic counseling, the neurologic exam, the psychological/psychiatric interview, testing, and continued counseling.

In the pretesting genetic counseling step, the patient obtains basic background knowledge about genetics, how Huntington disease is inherited, and the testing procedure.

The purpose of the neurologic exam is to look for signs and symptoms of the disorder. Throughout the neurologic exam, the patient's body movements, eye movements, hearing, and balance are assessed. Brain imaging scans may be used to look for structural changes.

The psychological/psychiatric interview includes an assessment of the patient's mental and emotional state by a mental health professional.

After the completion of these preliminary steps, the patient will wait for a predetermined amount of time before undergoing testing so that he or she can make an informed decision.

Juvenile Huntington disease

Juvenile Huntington disease is seen in individuals younger than age 20. Ten percent of all Huntington disease cases are children or adolescents, according to the HDSA. The gene mutation resulting in the repetition of the CAG base pairs is present, but the repeats will be in larger numbers than what's seen in a patient who develops signs and symptoms as an adult. After a juvenile is diagnosed with Huntington disease, the average life expectancy is 15 years; juvenile Huntington disease may progress at a faster pace than adult onset.

The signs and symptoms seen in juvenile Huntington disease may differ from those seen in adult onset. Unlike adults with Huntington disease, juveniles typically present with either mild chorea or no involuntary movements at all. In many cases, individuals will be symptomatic for a number of years before diagnosis, reporting that they began to have difficulty with tasks, such as writing, sports, and so on. According to the HDSA and the Huntington Society of Canada, the following early signs and symptoms may be observed in juvenile Huntington disease:

- rigidity
- slowness and stiffness
- awkward walking
- diminished coordination
- personality changes
- behavior changes
- poor judgment
- slowed response time
- poor school performance
- difficulty learning new information
- inability or difficulty completing tasks previously learned.

These individuals may also have difficulty with speaking, chewing, and swallowing.

Diagnosing juvenile Huntington disease can be a challenge because this disorder is rare and not often seen. Obtaining a family history is a very important piece of the diagnostic puzzle, but can be challenging if the child or one of his or her parents was adopted because the family history may be unknown. In some instances, the parents may be unaware that Huntington disease runs in their family. As with the adult patient with Huntington disease, a definitive diagnosis is made with genetic testing. Before diagnosis, it isn't uncommon to find that the juvenile has been diagnosed with attention deficit disorder or labeled as having "behavior problems."

As with the adult patient with Huntington disease, the disorder is incurable; medical management is aimed at symptom control. Be aware that the loss of independence will be particularly challenging for these individuals. Even more challenging is the knowledge that the disease can't be cured. The juvenile with Huntington disease may feel a wide range of emotions, such as frustration, anger, grief, fear, sadness, and helplessness. In some instances, they may express negative emotions through aggressive behaviors.

Once the test has been completed, there are three potential outcomes: negative, positive, or uninformative. A negative test result means that the patient doesn't carry the genetic mutation and won't develop the disease. A positive result means that the patient carries the Huntington disease allele and will eventually develop the disease. An uninformative result provides results that are unclear; this result happens to only 1% to 4% of patients who are tested.

Complications

Patients with Huntington disease have increased risks associated with the disease. These patients may have impaired judgment, impulse control, and self-awareness, which can lead to putting themselves in unsafe situations. Potential risks include:

- choking
- aspiration
- malnourishment

- falls
- pressure injuries
- personal neglect
- impulsive behavior
- aggression to others
- inappropriate sexual behavior
- dangerous driving
- fire from a variety of sources, such as smoking and cooking
- suicide.

Patients with Huntington disease have a shorter life expectancy, often due to complications associated with the disease, and will usually die from pneumonia, heart disease, or aspiration. Other complications include gastrointestinal disease, such as pancreatic cancer; genitourinary diseases, such as kidney failure; and suicide. These patients may also experience chronic stress, which has been associated with hypertension, increased risk of myocardial infarction, and tumor growth.

Management

Because there's no cure for Huntington disease, treatment is aimed at controlling symptoms and minimizing complications. Medications for emotional and movement problems are often prescribed. In the early stages of the disease, medical treatment may be beneficial for depression and behavioral problems.

In the middle stage, medication for chorea may be beneficial, as well as occupational, physical, and speech therapy. Tetrabenazine was the first drug approved by the U.S. FDA for the treatment of Huntington disease. In 2008, this medication was approved for the treatment of chorea.

When caring for a patient with Huntington disease, monitor for medication adverse reactions, such as fatigue, restlessness, and hyperexcitability. Research has shown that individuals who exercise and are physically fit tend to have less debilitating symptoms than those who don't, so educate the patient about the importance of regular exercise. Due to the fact that choking and aspiration are potential complications, take extra care to prevent both of these in the patient with Huntington disease.

Addressing the risks associated with Huntington disease requires a multidisciplinary approach. Nursing should work with speech, physical, and occupational therapy; registered dietitians; primary care providers; and psychiatrists. As the disease progresses and the patient becomes more symptomatic, the level of risk increases.

Data have shown that patients with Huntington disease have higher suicide rates than the general population. This may be due to the progressive nature of the disease, as well as the associated stress. Be aware that suicide is most common in the earlier stages of the disorder when the patient is still cognitively able to carry out suicide.

Another factor that increases the risks for these patients is that they have a decreased ability to adjust to change. When caring for a patient with Huntington disease, you need to be able to prioritize risks so that appropriate interventions can be initiated.

Nursing specialty

Patients with Huntington disease have unique care requirements, highlighting the importance of nurses with specialized knowledge about the disorder. Specialist nurses have disease-, service-, and person-specific knowledge, enabling them to provide more appropriate and effective patient care. Research has shown that Huntington disease specialist nurses reduce hospital admissions by more than 50%, decrease the length of hospital stays by nearly half, and improve care quality in both inpatient and outpatient settings. In addition to keeping these patients healthier for longer, nurse specialists are also able to more effectively advocate for their patients.

These nurses typically spend a great deal of time with patients and their families, building a strong rapport and improving continuity. The nurse specialist is responsible for developing a detailed care plan and can provide beneficial education for patients, families, and other healthcare providers. More research needs to be conducted on the benefits and roles of the Huntington disease nurse specialist. If this is an area of interest to you, encourage your facility to consider the option.

Patient care

Nurses caring for patients with Huntington disease need a broad base of knowledge. For example, children of a parent with Huntington disease may experience tremendous stress resulting from either uncertainty about whether they'll develop the disease or the certainty that they will. It's also important to understand the fears that an individual may have regarding genetic testing. Some patients worry that genetic testing may result in discrimination at work and by insurance companies. Assess for these concerns and help address them. A thorough understanding of the ethical concerns surrounding genetic testing will help you provide the best care and support for your patient.

When working with juveniles with Huntington disease, your role will vary, depending on the child's response to the diagnosis, disease, and symptoms. Typically, you'll want to encourage the family and the child's school to allow him or her to be as independent as possible for as long as possible. It's also important to encourage friendships and new experiences, and keep the child involved and educated about his or her care plan. Provide the family with information about support services. The family may experience both emotional and financial problems after a diagnosis of juvenile Huntington disease. There may come a point when one parent needs to stay home with the child to provide care, losing income. Additionally, it's essential to be aware of the stress any other children in the house may be feeling. These children may be afraid that they'll get the disease or resentful of the attention the child with the disease is getting. Being knowledgeable about the support services available is tremendously helpful for these families.

When working with adult patients in the healthcare setting, ask family members and/or caregivers about the patient's routine. As discussed previously, patients with Huntington disease have a difficult time adapting to change; developing a care plan based on the patient's routine helps alleviate complications associated with changes. It may also be helpful to ask family members and/or caregivers to bring in objects from home to make the patient feel more comfortable. Due to the fact that safety can be a concern, modify the environment to keep the patient safe. As with all patient rooms, a clutter-free environment should be maintained, the bed should be in the low position with the brakes locked, and appropriate lighting should be utilized. Keep in mind that walking can pose a safety risk for the patient and the person walking with the patient. Because impulsivity and poor judgment can occur with this disorder, the patient should be roomed near the nurses' station so frequent checks can be performed. Lastly, it's important to discuss end-of-life issues with the patient while he or she is still able.

Research has been conducted regarding the benefits of Huntington disease nurse specialists. For more information, see *Nursing specialty.*

on the web

Huntington Society of Canada: https://www.huntingtonsociety.ca

Huntington's Disease Society of America: www.hdsa.org

Mayo Clinic: www.mayoclinic.org/diseases-conditions/huntingtons-disease/basics/definition/con-20030685

National Institute of Neurological Disorders and Stroke: https://www.ninds.nih.gov/Disorders/All-Disorders/Huntingtons-Disease-Information-Page

Knowledge zone

It's important to be knowledgeable about Huntington disease because it's

progressive, incurable, and can be seen in children as well as adults. Understanding this disorder will lead to improved patient safety and care quality. ■

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Amanda Perkins is an Assistant Professor of Nursing at Vermont Tech in Randolph, Vt., and a *Nursing made Incredibly Easy!* Editorial Board Member.

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