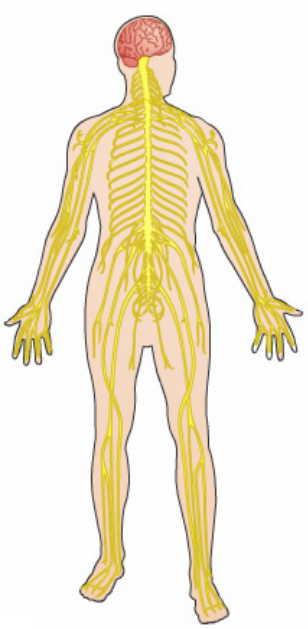
Huntington's Disease

Introduction

Huntington's disease is a progressive brain disorder that causes uncontrolled movements, emotional problems, and loss of thinking ability.

Huntington's disease is inherited. This means that the defective gene that causes Huntington's disease is passed on from parent to child. If one of your parents has Huntington's disease, you have a 50-50 chance of getting it.

This reference summary helps you understand Huntington's disease. It covers the symptoms, causes, diagnosis and treatment.



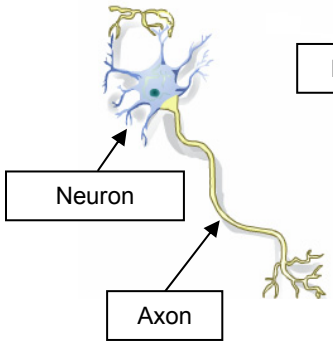
Nervous System

The Central Nervous System

The brain and spinal cord are called the central nervous system. The nerves in the body, such as the arms, legs, chest, abdomen, and pelvis, make up the peripheral nervous system.

The brain is similar to a very complex computer that processes input from our senses and then tells the body how to respond, such as by talking or moving.

The main cells that make up the brain and spinal cord are called neurons. The neurons are the thinking cells of the brain. They communicate with each other by sending electric signals through wires called axons.



There are three types of neurons:

1. Sensory neurons
2. Motor neurons
3. Interneurons

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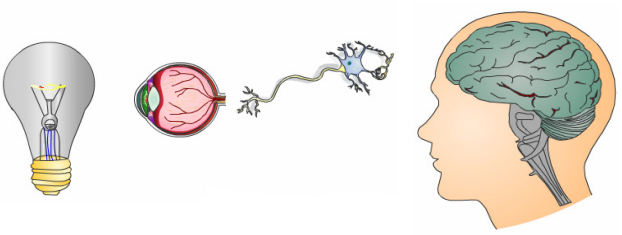
The sensory neurons carry information from the sense organs to the brain. Some examples of sense organs include the eyes and ears. Motor neurons receive and send messages from the body to the brain and back to the body. These messages control the muscles and glands of the body.



Sensory Neuron Carrying Information to the Brain

Interneurons communicate only within their immediate area in the brain.

Everything we think and do would be impossible without neurons. We need neurons to learn, talk and move.

Huntington's Disease

Huntington's disease causes certain neurons in the brain to waste away. The disease attacks neurons mainly in the basal ganglia.

The basal ganglia are a group of brain neurons deep in the brain, close to the thalamus. The thalamus is an area of the brain that helps process information from the senses. It also transmits this information to other parts of the brain.

The basal ganglia control emotion, movement and cognitive ability. Cognitive ability includes learning, thinking and problem-solving.

Because Huntington's disease causes the neurons in the basal ganglia to die, messages cannot be sent to or from this part of the brain. This means that Huntington's disease can cause uncontrolled movements, emotional problems, and loss of thinking ability.

Symptoms

People with Huntington's disease are born with the defective gene that causes Huntington's disease. However, symptoms usually don't appear until middle age.

Often the symptoms of Huntington's disease start when a person is in their 40s or 50s. Sometimes it starts earlier or later. When symptoms start earlier, the disease usually progresses faster.

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Early symptoms of Huntington's disease affect movement, including uncontrolled movements, clumsiness or balance problems.

The most common movement problems caused by Huntington's disease are called chorea and dystonia. Chorea is involuntary, jerky movements. Dystonia is involuntary muscle contractions which can cause twisting or repetitive motions.

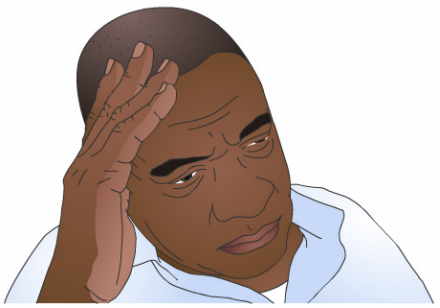
As Huntington's disease progresses, it can cause problems with walking, talking or swallowing. Other common movement problems seen in people with Huntington's disease include:

- Abnormal eye movements
- Firm, tense muscles
- Poor posture

Huntington's disease can also affect a person's emotions and cognitive ability late in the disease. Some people stop recognizing family members or have a difficult time expressing emotions.

If a person's emotions are affected, Huntington's disease often causes depression. Depression is when a person is sad all the time. This sadness can interfere with life at home and work.

Other symptoms of depression include:



- Changes in appetite
- Feeling worthless
- Little or no interest in normal activities or hobbies
- Shying away from family and friends
- Sleeping more than normal or having difficulty sleeping
- Tiredness
- Thoughts of death or suicide

Other mental illnesses that can also be caused by Huntington's disease include obsessive-compulsive disorder or bipolar disorder. These disorders can also cause changes in mood or personality.

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Obsessive-compulsive disorder, or OCD, is characterized by repeated, upsetting thoughts called obsessions. These obsessions cause a person with OCD to do the same action over and over to try to make the thoughts go away. These repeated actions are called compulsions.

Bipolar disorder is a mental illness that causes dramatic mood swings. People with bipolar disorder may go from an overly energetic, high feeling called mania, to a sad and hopeless feeling called depression.

Huntington's disease can also result in the loss of cognitive ability. This may cause the following symptoms:

- Acting without thinking
- Difficulty focusing or learning new information
- Difficulty starting a task or conversation
- Perseveration, or the tendency to get stuck on a thought or action
- Problems with spatial perception, which is the ability to see and evaluate how objects are located in the surrounding space or environment
- Trouble finding the right words
- Trouble planning or organizing tasks

Causes

Huntington's disease is inherited. This means that the defective gene that causes Huntington's disease is passed on from parent to child. The sons and daughters of a parent who has Huntington's disease each have a 50% chance of inheriting the gene that causes Huntington's disease.

Rarely, some people may develop Huntington's disease without having a family history of it. This is due to a genetic change that happens during the development of the father's sperm.

This section explains more about how HD genes are inherited for those who are interested.

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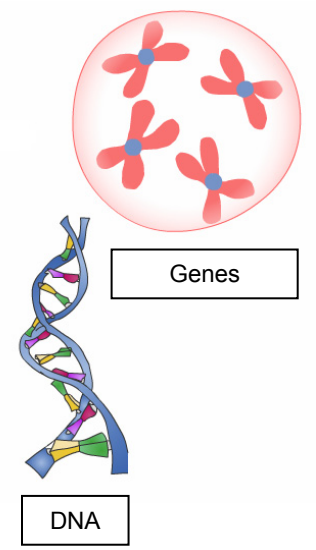
Genes tell the body to make certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have a lot to do with our health.



Each person has 23 pairs of chromosomes. Genes are found on chromosomes. The gene linked to Huntington's disease is on chromosome 4. It is called the HTT gene. The HTT gene makes a protein called huntingtin. The exact purpose of this protein is unknown. However, it plays a role in the brain's neurons.

The HTT gene either works correctly or it is defective. We will call the good HTT gene that works correctly "g" for good. We will call the defective HTT gene that does not work correctly "D" for defective.

Everybody has 2 HTT genes: 1 from the father and 1 from the mother. If one of the two genes is defective, the person will likely develop Huntington's disease. Here is a summary:

- gD has HD because one gene is D or defective
- gg has two good genes and does not have HD
- Dg has a defective gene and has HD
- DD has two defective genes and has HD

If both parents are gg, their child will not have the defective gene that leads to Huntington's disease. Each parent can only give one good HTT gene.

If only one parent is gD, meaning they have the defective HTT gene, their child has a 50-50 chance of getting it. The parent may pass on the good gene or they may pass on the defective gene.

If a child does not inherit the Huntington's disease gene, he or she will not develop the disease and cannot pass it on to his or her children. If a person inherits the defective gene, he or she will most likely develop the disease sooner or later and may pass it on to his or her children.

Depending on their genetic makeup, some people who inherit a defective HTT gene may not develop Huntington's disease but are still able to pass it on to their children. Others may develop the disease earlier in life, meaning the disease will progress faster.

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In some families, all children may inherit the Huntington's disease gene. In other families, none do. If one child has the defective gene, it does not affect whether the other children will also inherit it.

Diagnosis

Your healthcare provider will ask you about your symptoms. He or she will also perform a physical exam.

Your healthcare provider may also request neurological and psychiatric exams. A neurological exam checks your muscle strength and how well your nerves are working. A psychiatric exam evaluates your mental health.

Some tests may be ordered to check the brain. Brain imaging tests can show if certain structures in the brain have been affected by Huntington's disease. These tests can also rule out other causes of your symptoms.

An electroencephalogram, or EEG for short, may also be done. An EEG records the electrical activity of the brain.

A blood test can confirm the diagnosis. It can tell if you have the defective gene that causes Huntington's disease. If you have the gene, you will likely develop the disease.

If you have a family history of Huntington's disease, you may consider being tested for the defective gene before you have any symptoms. This is called predictive testing. Genetic counseling can help you weigh the risks and benefits of taking the test.

Treatment

There is no cure for Huntington's disease. Medicines can help manage some of the symptoms, but cannot slow down or stop the disease.

Medications are available to help treat movement problems caused by Huntington's disease. For example, tetrabenazine (Xenazine®) is specifically used to treat the involuntary, jerking movements caused by Huntington's disease.

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Antipsychotic drugs and other medicines can also help suppress involuntary movements. Emotional problems caused by Huntington's disease may also be treated with medications. The medications used depend on the specific problem.

Some medications used to treat emotional problems include:

- Antidepressants
- Antipsychotic drugs
- Mood-stabilizing drugs

The symptoms of Huntington's disease may also be treated with different types of therapy. For example, psychotherapy is a form of talk therapy. It can help a person with Huntington's disease cope and adjust to life as the symptoms progress.

Speech therapy may also be beneficial for people who have trouble talking or swallowing. Speech therapists can help improve communication and strengthen muscles in the throat.

Physical therapy focuses on exercises that improve strength, flexibility and balance. Using these exercises may help with the movement problems caused by Huntington's disease.

Occupational therapy can also be used to treat the symptoms of Huntington's disease. An occupational therapist can introduce devices that aid movement and improve independence.

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