Friedreich's Ataxia

Introduction

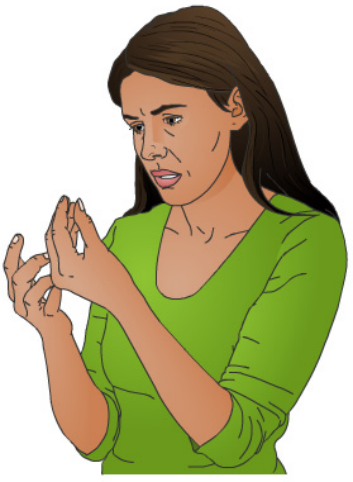
Friedreich's ataxia is a disease that damages your nervous system. It affects your spinal cord and the nerves that control muscle movement in your arms and legs. The main symptom is ataxia. Ataxia means trouble controlling movements. The symptoms of FA can be treated with medicines, braces, surgery and physical therapy.

This reference summary explains Friedreich's ataxia. It covers symptoms and causes of the disease. It also talks about diagnosis and treatment.

The Nervous System

Together, the brain and spinal cord are called the "central nervous system." The nerves in the rest of the body are called the "peripheral nervous system."

The brain is the command center of the body. It lets us think and feel. It also receives information and sends orders to different parts of the body. Orders from the brain travel through the spinal cord. From the spinal cord, orders travel to the rest of the body through peripheral nerves.



Healthy nerves send and receive messages about movement. For example, if you want to raise your hand, your brain tells your arm and hand to do it. Healthy nerves also send and receive messages about sensation, or feeling. If you burn your hand while cooking, the nerves in the skin send pain signals to your brain. If nerves become damaged, they cannot send and receive messages well. This can cause problems with movement and sensation. Nerve damage can happen anywhere in the body. Important organs can also be affected by nerve problems.

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Friedreich's Ataxia

Friedreich's ataxia is also called FA or FRDA. It is a rare inherited disease. This means it is passed from parent to child. It causes nervous system damage and movement problems. FA usually begins in childhood. It leads to impaired muscle coordination that gets worse over time.

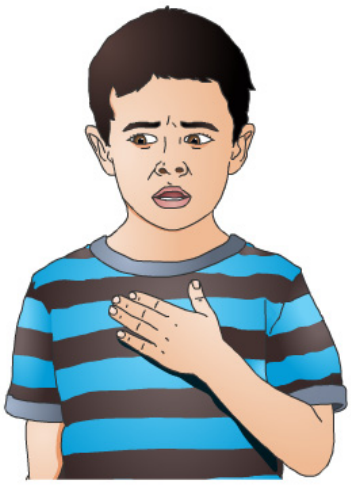
In FA, the spinal cord and peripheral nerves become thinner. The cerebellum is also affected. The cerebellum is the part of the brain that helps control balance and movement. Damage to the nerves and brain causes awkward, unsteady movements. It also causes problems with sensations. In some cases, FA causes problems in the heart and spine. Some people with the condition may develop diabetes. FA does not affect thinking and reasoning abilities.

Symptoms

Symptoms of FA usually begin between the ages of 5 and 15. The main symptom is ataxia, which means trouble controlling movements.

Other symptoms of FA include:

- Difficulty walking.
- Involuntary eye movements.
- Muscle weakness.
- Scoliosis, which is curving of the spine to one side.
- Speech problems.



FA can also affect the heart, resulting in:

- Chest pain.
- Heart palpitations.
- Shortness of breath.

Most individuals with FA get tired easily. They may also find that they take a longer time to recover from common illnesses like colds and the flu.

People with FA usually need a wheelchair 10 to 20 years after symptoms first appear. But many people with the disease can live normal lives.

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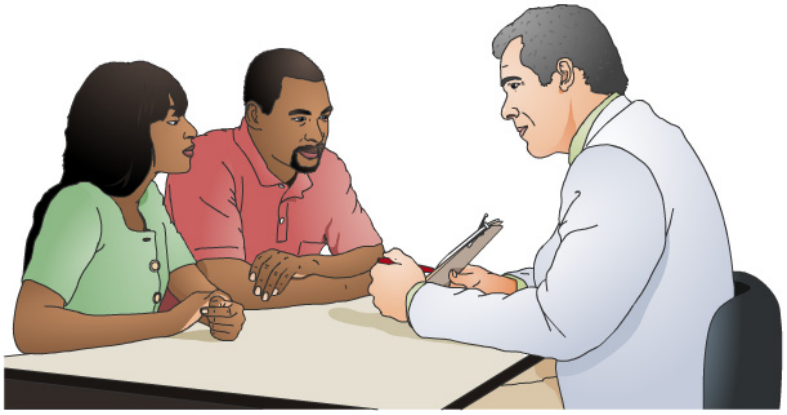
In severe cases of FA, people become incapacitated. They can no longer care for themselves and must rely on others for daily living.

Causes

FA is inherited. This means it is passed on from parent to child. It is caused by a defective, or abnormal, gene. FA is not contagious. You cannot catch it from another person.

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. FA is caused by a mutation, or change, in a gene labeled FXN. The FXN gene controls the production of a protein called "frataxin."

Frataxin is found in the parts of the cell that make energy. If the body does not have enough frataxin, it cannot make energy. This can lead to "oxidative stress." Oxidative stress is when there is a buildup of harmful toxins and free radicals in the cells. Free radicals can destroy cells and can harm the body.



Certain cells are affected by the lack of frataxin. These include cells in the:

- Brain.
- Heart muscle.
- Peripheral nerves.
- Spinal cord.

Damage to these cells results in the symptoms of FA. FA only happens in someone who inherits two defective copies of the gene, one from each parent. A person who has only one abnormal copy of the gene is called a carrier. A carrier will not develop FA. But he or she can pass the gene mutation on to his or her children. If both parents are carriers, their children will have a 1 in 4 chance of having the disease. Their children will also have a 1 in 2 chance of inheriting one abnormal gene and being carriers.

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Diagnosis

A health care provider will first ask about your child's medical history and symptoms. He or she will also ask about family medical history.

A physical exam will be performed. The health care provider will check for:

- Balance problems.
- Lack of reflexes.
- Loss of sensation in the joints.
- Signs of neurological problems.

A genetic blood test is used to diagnose FA. It shows if your child has two defective FXN genes.

Other tests may also be done to check for damage caused by FA. These include:

- CT scan.
- Echocardiogram.
- Electrocardiogram, or ECG.
- Electromyogram, or EMG.
- MRI.
- Nerve conduction studies.

A CT scan is an x-ray machine linked to a computer. A CT scan takes a series of detailed pictures of your organs. You may be given contrast material by mouth or injection to make abnormal areas easier to see.

An echocardiogram uses sound waves to create moving pictures of the heart. An ECG uses electrodes attached to the chest. The ECG creates a graph of the electrical activity, or beat pattern, of the heart. An EMG uses electrodes to measure the electrical activity of muscle cells. An MRI uses strong magnets to create images of the inside of the body. Nerve conduction studies measure the speed with which nerves transmit signals.

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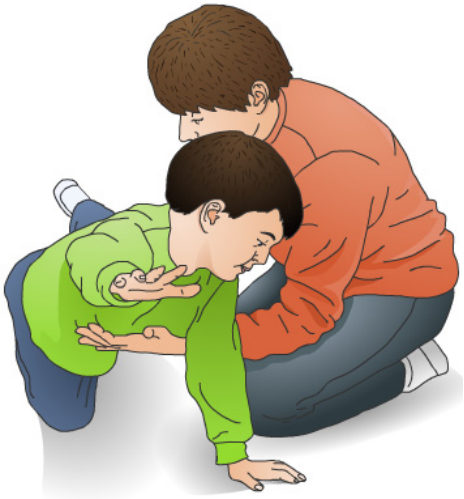
Treatment

There is no cure for FA. Treatment focuses on relieving symptoms of the disease. Treatment may include:

- Medicines.
- Braces.
- Surgery.
- Physical therapy.
- Speech therapy.
- Assistive devices.



Medicines can be used to treat diabetes or heart problems caused by FA. Foot deformities and scoliosis can be corrected with braces or surgery. Physical therapy may prolong use of the arms and legs. Speech therapists can help retrain muscles in the face and neck. This can improve speech and swallowing. Assistive devices, such as a wheelchair or walker, can help a person with FA get around. Other devices can help with daily activities.

Summary

Friedreich's ataxia, or FA, is a disease that damages your nervous system. It affects your spinal cord and the nerves that control muscle movement in your arms and legs. The main symptom is ataxia. Ataxia means trouble controlling movements.

Damage to the nerves and brain in people with FA causes awkward, unsteady movements. It also causes problems with sensations. In some cases, FA affects the heart and spine. Some people with the condition may develop diabetes.

FA is inherited. This means it is passed on from parent to child. FA is caused by a mutation, or change, in a gene labeled FXN. The FXN gene controls the production of a protein called "frataxin." If the body does not have enough frataxin, it cannot make energy. A genetic blood test is used to diagnose FA. It shows if your child has two defective FXN genes. Other tests may also be done to check for damage caused by FA. Thankfully, lifelong treatment can help relieve many of the symptoms of FA.

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