# Friedreich's Ataxia

Friedreich's ataxia is an inherited disease that causes progressive damage to the nervous system. Loss of muscle coordination (*ataxia*) results from the degeneration of nerve tissue in the spinal cord and of nerves that control muscle movement in the arms and legs.

## SYMPTOMS

Symptoms range from muscle weakness and speech problems to heart disease. Symptoms usually begin between the ages of 5 and 15. They can also appear as early as 18 months or as late as 30 years of age. The first symptom is usually difficulty in walking. The ataxia gradually worsens and slowly spreads to the arms and then the trunk. Foot deformities such as clubfoot, flexion (involuntary bending) of the toes, hammer toes, or foot inversion (turning in) may be early signs. Rapid, rhythmic, involuntary movements of the eyeball are common. Most people with Friedreich's ataxia develop curving of the spine to one side (*scoliosis*). If severe, scoliosis may impair breathing. Other symptoms include chest pain, shortness of breath, and heart palpitations (irregular heartbeat).

## DIAGNOSIS

Doctors diagnose Friedreich's ataxia by careful clinical examination. This includes a medical history and a thorough physical examination. Several tests may be performed, including electromyogram (EMG) and genetic testing.

## TREATMENT

There is currently no cure or treatment for Friedreich's ataxia. However, many of the symptoms and complications can be treated. This can help patients maintain optimal functioning as long as possible. Diabetes and heart problems can be treated with medications. Orthopedic problems such as foot deformities and scoliosis can be treated with braces or surgery. Physical therapy may prolong use of the arms and legs.

## WHAT WILL HAPPEN

Generally, within 15 to 20 years after the appearance of the first symptoms, the person is confined to a wheelchair, and in later stages of the disease, individuals become completely incapacitated. Most people with Friedreich's ataxia die in early adulthood if there is significant heart disease, the most common cause of death. Those with less severe symptoms tend to live much longer.

## RESEARCH BEING DONE

Studies have revealed that frataxin, a protein, is severely reduced in patients with Friedreich's ataxia. This protein is normally present in the nervous system, the heart, and the pancreas. Studies have shown that patients have abnormally high levels of iron in their heart tissue. It is believed that the nervous system, heart, and pancreas may be particularly susceptible to damage from free radicals (produced when the excess iron reacts with oxygen). Once certain cells in these tissues are destroyed by free radicals they cannot be replaced. Nerve and muscle cells also have metabolic needs that may make them vulnerable to free radical damage. The discovery of the genetic mutation that causes Friedreich's ataxia has added new impetus to research efforts on this disease.

**Research is continually being done to learn more about this problem.**

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