CONDITION

Friedreich ataxia

Friedreich ataxia is an inherited condition that progressively affects coordination, movement, sensation, and often the heart.

WHAT IT IS
Inherited progressive condition affecting coordination, sensation, and the heart

BODY SYSTEM
Nervous system, muscles, heart and circulation, pancreas

MAIN SYMPTOMS
Poor balance, unsteady walking, weakness, slurred speech

WHAT CAUSES IT
Inherited changes affecting the frataxin protein

HOW IT IS DIAGNOSED
Genetic testing for changes in the FXN gene

MAIN TREATMENT
Progression-slowing medicine, rehabilitation, and complication management

CAN IT BE CURED?
No - treatment supports function and may slow progression

IS IT AN EMERGENCY?
Sometimes - chest pain, severe breathlessness, or fainting

LONG-TERM OUTLOOK
Progressive disability, with outlook strongly influenced by heart involvement

Overview

Friedreich ataxia is a rare, inherited condition that gradually damages parts of the nervous system. "Ataxia" means difficulty coordinating movement. The condition can affect walking, balance, speech, sensation, and hand control.

It can also affect other organs. Many people develop heart muscle changes, and some develop diabetes, curvature of the spine, hearing loss, or vision problems.

Symptoms often begin during childhood or the teenage years, but may start earlier or in adulthood. Later-onset Friedreich ataxia may progress more slowly, although each person's course is different.

Friedreich ataxia is progressive, meaning its effects usually increase over time. It does not

directly affect intelligence. People may need more physical support as it advances, but they continue to learn, work, make decisions, and participate in family and community life.

There is no cure. Treatment can help maintain mobility and independence, manage complications, and support emotional well-being. A medicine intended to slow progression is available to some people in some countries.

Symptoms

Early signs often include difficulty running, frequent tripping, poor balance, or an unusual walking pattern. These may initially be mistaken for clumsiness.

Symptoms usually affect both sides of the body and may include:

Leg reflexes are often reduced or absent. Nerve damage can also make it harder to know where the feet and legs are without looking.

Heart involvement may cause breathlessness, reduced exercise tolerance, palpitations, chest discomfort, dizziness, or fainting. Heart changes can be present before symptoms appear.

Some people develop diabetes. Possible signs include increased thirst, frequent urination, tiredness, blurred vision, or unexplained weight loss.

- Unsteady walking and poor balance
- Reduced coordination of the arms and hands
- Muscle weakness, stiffness, cramps, or tiredness
- Reduced sensation, especially awareness of limb position
- Slurred or slowed speech
- Difficulty swallowing
- Abnormal eye movements or reduced vision
- Hearing loss or difficulty understanding speech in noise
- Foot changes, such as high arches
- Scoliosis, meaning a sideways curve of the spine

Causes and risk factors

Friedreich ataxia is caused by harmful changes in the FXN gene. This gene provides instructions for making frataxin, a protein that helps cells produce energy and manage iron

safely. Low frataxin levels particularly affect nerve cells, heart muscle, and other tissues with high energy needs.

Most people have an unusually repeated section of DNA, called a GAA repeat expansion, in both copies of the FXN gene. A smaller number have an expansion in one copy and a different harmful FXN change in the other.

Friedreich ataxia has an autosomal recessive inheritance pattern. A person usually inherits one altered FXN copy from each parent. The parents are commonly healthy carriers because each has one working copy.

When both parents are carriers, each pregnancy has:

These chances apply separately to every pregnancy. The repeat length can influence when symptoms begin and how quickly they progress, but cannot predict an individual course with certainty.

- A 1 in 4 chance of a child having Friedreich ataxia
- A 1 in 2 chance of a child being an unaffected carrier
- A 1 in 4 chance of a child inheriting neither altered copy

Diagnosis

A healthcare professional may suspect Friedreich ataxia from balance problems, reduced reflexes, sensory loss, speech changes, or heart findings. Age at symptom onset and family history may provide clues, although many people have no known family history because carriers usually have no symptoms.

A blood or saliva genetic test confirms the diagnosis. Testing first looks for GAA repeat expansions in the FXN gene. If only one expansion is found, additional sequencing may look for another harmful FXN change.

Genetic counselling can explain the result, its implications for relatives, and the limits of genetic predictions. Testing relatives should be a personal, informed decision.

Other assessments show how the condition is affecting the body and may include:

MRI scans and nerve tests may help rule out other causes of ataxia. A negative FXN test may lead to testing for other inherited or acquired conditions.

- An electrocardiogram and heart imaging
- Blood glucose or other diabetes tests

- Hearing and vision assessments
- Spine, mobility, speech, and swallowing assessments
- Blood tests before and during certain treatments

Treatment and management

Care is usually coordinated across neurology, cardiology, rehabilitation, and other specialties. The plan changes as needs change.

Omaveloxolone is intended to slow neurological progression. In the United States, it is approved for people with Friedreich ataxia aged 16 years and older. Approval and availability differ between countries. It is not a cure and does not reverse established nerve damage.

Omaveloxolone can affect liver test results, cholesterol levels, and other measurements, so blood tests are needed before and during treatment. It can interact with other medicines and may make hormonal contraceptives less effective. A prescribing clinician or pharmacist can explain the precautions that apply to you.

Physical therapy may support strength, flexibility, balance, transfers, and wheelchair skills. Occupational therapy can recommend equipment or adaptations for school, work, personal care, cooking, writing, and computer use.

Speech and language therapy can support communication and swallowing. Communication devices may help if speech becomes difficult to understand. A swallowing assessment can guide food textures or eating strategies when choking or weight loss is a concern.

Treatment for complications may include:

Regular exercise adapted to your abilities may help maintain fitness and participation. It should account for balance, heart health, fatigue, and fall risk.

- Heart medicines for rhythm problems or weakened heart function
- Diabetes monitoring and treatment
- Bracing, therapy, or surgery for significant scoliosis
- Hearing aids or other hearing technology
- Mobility aids, orthotics, wheelchairs, and home adaptations
- Psychological support for anxiety, low mood, grief, or adjustment

Complications

Increasing difficulty with walking and transfers can lead to falls, reduced activity, joint stiffness, pressure injuries, and loss of independence if equipment and support do not keep pace with changing needs.

Heart muscle thickening, called hypertrophic cardiomyopathy, is common. Some people later develop weakened pumping function, heart failure, or abnormal rhythms. Heart involvement strongly influences long-term health, so routine cardiac follow-up is important even without symptoms.

Severe scoliosis may cause pain, altered posture, or breathing difficulties. Foot changes can affect comfort and footwear.

Swallowing problems may cause weight loss, dehydration, or aspiration, in which food enters the airway and may cause a chest infection. Poorly controlled diabetes can damage blood vessels, kidneys, eyes, and nerves.

Hearing and vision changes may affect communication, education, work, and navigation. Prompt assessment can identify useful supports.

Prevention

Friedreich ataxia cannot be prevented in someone who has inherited the FXN changes that cause it. Lifestyle choices do not cause the condition, and parents do not cause it through anything they did during pregnancy.

Genetic counselling can explain carrier testing and reproductive options for people with a family history or confirmed FXN change. Options may include testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices, not requirements.

Complications cannot always be prevented, but regular heart, diabetes, mobility, hearing, vision, and swallowing assessments can identify problems earlier. Vaccination, dental care, safe mobility equipment, and appropriate physical activity support general health.

Living with it and what to expect

Friedreich ataxia usually changes gradually over years. Balance and walking often become harder first. Hand coordination, speech, swallowing, hearing, vision, and strength may change later. Heart problems and diabetes may follow a different timeline from neurological

symptoms.

Many people eventually use a wheelchair for some or all mobility. The timing cannot be predicted from age or genetic results alone. Mobility equipment can reduce falls, conserve energy, and make education, work, travel, and social activities more accessible.

Equipment should be reviewed as posture, strength, and daily activities change. Planning for accessible housing, transport, school, or work can reduce disruption.

People with Friedreich ataxia generally retain their thinking and understanding abilities. Fatigue, hearing loss, slow speech, or physical limitations can be mistaken for impaired comprehension. Allowing enough time and using accessible communication methods can help.

Life expectancy may be shortened, particularly with serious heart complications. Some people live well into later adulthood, especially with later-onset symptoms or less severe heart involvement. No genetic test or symptom pattern can provide an exact forecast.

Counsellors, peer groups, disability services, and the wider Friedreich ataxia community can help with adjustment, relationships, education, work, and future planning.

When to get medical help

Arrange routine follow-up with your care team even if symptoms seem stable. Heart changes and diabetes may not cause early warning signs.

Request a prompt medical review for new or worsening:

Seek emergency help for chest pain, severe difficulty breathing, fainting, or a sustained fast or irregular heartbeat with dizziness. Severe confusion, repeated vomiting, deep rapid breathing, or marked drowsiness alongside symptoms of high blood sugar also need emergency assessment.

A sudden change is not always part of Friedreich ataxia. New symptoms may have another cause requiring separate treatment.

- Falls, weakness, pain, or loss of mobility
- Palpitations, breathlessness, ankle swelling, or reduced exercise tolerance
- Choking, coughing during meals, weight loss, or repeated chest infections
- Increased thirst, frequent urination, or unexplained weight change
- Hearing, vision, mood, or sleep problems

Common questions

Is Friedreich ataxia the same as multiple sclerosis?

No. Friedreich ataxia is inherited and caused by changes in the FXN gene. Multiple sclerosis involves immune-system damage to the brain and spinal cord, although some movement symptoms may appear similar.

Can Friedreich ataxia be cured?

There is no cure. Omaveloxolone may slow progression for some eligible people, while rehabilitation and treatment of complications help protect health and independence.

Does Friedreich ataxia affect intelligence?

It does not usually reduce intelligence. Speech changes, hearing loss, fatigue, or slow movement may make communication take longer without affecting understanding.

Will I need a wheelchair?

Many people eventually use one, but when and how often varies. Some initially use a wheelchair for long distances or busy days. Early equipment assessment can improve safety and conserve energy.

Can someone with Friedreich ataxia have children?

Yes. Fertility is not necessarily affected. A person with Friedreich ataxia passes an altered FXN copy to each child, but whether the child develops the condition also depends on the other biological parent's FXN genes. Genetic counselling can clarify the chances.

Is Friedreich ataxia painful?

Pain is not always a main feature, but muscle cramps, back or joint pain, nerve pain, or discomfort from scoliosis and foot changes can occur. New or persistent pain should be assessed.

Sources

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