
Friedreich's ataxia

Summary

- Friedreich's ataxia is a relatively rare inherited disease of the nervous system characterised by the gradual loss of coordination.
 - The symptoms of Friedreich's ataxia are caused by the gradual deterioration of nerve cells (neurones) in the spinal cord and brain.
 - There is no cure, but some of the symptoms can be managed with medication and physical therapy.
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Friedreich's ataxia (also known as 'Friedreich') is a relatively rare inherited condition of the nervous system characterised by the gradual loss of coordination.

A person with Friedreich's ataxia has increasing difficulty with coordination, leading to an unsteady gait and slurred speech.

What causes Friedreich's ataxia?

Friedreich's ataxia is caused by changes in a gene called *FXN*, which is located on chromosome 9. This altered gene interferes with the production of a protein called frataxin.

Friedreich's ataxia is inherited in an autosomal recessive pattern, meaning both parents have one altered gene, but don't exhibit any symptoms themselves, since they have a second healthy copy. The children of parents who are both healthy carriers of a single altered *FXN* gene each have a 25 per cent chance of being affected by Friedreich's ataxia and a 50 per cent chance of being carriers like their parents.

Genetic counselling is recommended for parents who already have one child with Friedreich's ataxia, as well as relatives of people with the condition.

Symptoms of Friedreich's ataxia

The symptoms of Friedreich's ataxia are caused by the gradual deterioration of the neurones in the cerebellum and spinal cord, which prevents electrical messages from passing smoothly between neurones and muscles, leading to poor coordination and movement.

Friedreich's ataxia progresses slowly. At first, the person's lack of coordination may be mistaken for clumsiness.

The onset of symptoms usually occurs on average between 10 and 15 years of age, but can be as early as four years or as late as middle age. Most people with Friedreich's ataxia ultimately require the use of a wheelchair full-time, on average 15 years after symptom onset. Their life span may also be reduced. However, at this stage, medical technology cannot predict how fast the disease will progress – some people will progress faster than others.

Symptoms of Friedreich's ataxia include:

- lack of physical coordination
 - loss of balance
 - unsteadiness on the feet
 - the need to maintain balance by positioning the feet widely apart
 - unsteadiness is worsened if the person can't see (such as in the dark)
 - muscle weakness as the condition gets worse
 - walking abnormalities
 - lack of tendon reflexes in the legs
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- jerky movements and lack of coordination ultimately involving the arms, legs and body
- speech difficulties, such as slurring
- swallowing problems and an associated risk of choking
- increased susceptibility to respiratory infections
- numbness
- other abnormal changes in skin sensation, such as reduced ability to feel temperature, pressure or pain
- spine curvatures, such as **scoliosis** (where the spine curves sideways).

Complications of Friedreich's ataxia

Two out of three people with Friedreich's ataxia will have associated heart problems called cardiomyopathy. The heart muscle fibres degenerate and, ultimately, impair the heart's functioning. This can cause a range of problems, including palpitations and congestive heart failure.

Some of the other complications include:

- diabetes
- vision problems, which rarely include blindness
- hearing difficulties, including deafness. Many people with Friedreich's ataxia have normal hearing in a quiet environment, but significant issues hearing in a noisy environment.

Diagnosis of Friedreich's ataxia

Friedreich's ataxia is diagnosed using a number of tests including:

- medical history
- physical examination
- electrical nerve conduction tests
- **electrocardiogram (ECG)**
- echocardiogram (ultrasound of the heart)
- **x-rays**
- **magnetic resonance imaging (MRI)**
- genetic testing – the method for definitive diagnosis of Friedreich's ataxia.

Treatment for Friedreich's ataxia

There is currently no cure for Friedreich's ataxia. Treatment aims to ease some of the symptoms and may include:

- exercise, such as aquatic physical therapy to prolong the ability to walk and to help with balance, flexibility and accuracy of limb movements
- regular heart monitoring
- regular assessment of ataxia and muscle weakness to enable the appropriate referral to the required specialists
- speech and language pathology to assist with speech difficulties
- maintaining a healthy weight for your height
- orthopaedic aids
- protective foot care
- adaptations to the home, such as wheelchair access ramps
- physiotherapy
- medication to reduce muscle tremors and spasms
- medication to treat heart palpitations
- medication, such as insulin, to treat diabetes
- surgery to correct abnormal spine curvatures.

Research into the function of the deficient protein, frataxin, may offer potential for future beneficial treatments.

Genetic counselling and Friedreich's ataxia

If your child or another family member has been diagnosed with Friedreich's ataxia, or if it runs in your family, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Friedreich's ataxia and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If Friedreich's ataxia runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

Victorian Clinical Genetics Services (VCGS) provides genetic consultation, counselling, testing and diagnostic services for children, adults, families, and prospective parents.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Friedreich's ataxia.

Where to get help

- Your **GP (doctor)**
- **Muscular Dystrophy Australia** Tel. **(03) 9320 9555**
- **fara (Friedreich Ataxia Research Association)** Tel. **(03) 9818 6551**
- **Genetic Support Network of Victoria (GSNV)** Tel. **(03) 8341 6315**
- **Victorian Clinical Genetics Services (VCGS)** Tel. **1300 118 247**

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