

Friedreich ataxia

Description

Friedreich ataxia is a genetic condition that affects the nervous system and causes movement problems. People with this condition develop impaired muscle coordination (ataxia) that worsens over time. Other features of this condition include the gradual loss of strength and sensation in the arms and legs; muscle stiffness (spasticity); and impaired speech, hearing, and vision. Individuals with Friedreich ataxia often have a form of heart disease called hypertrophic cardiomyopathy, which enlarges and weakens the heart muscle and can be life-threatening. Some affected individuals develop diabetes or an abnormal curvature of the spine (scoliosis).

Most people with Friedreich ataxia begin to experience the signs and symptoms of the disorder between ages 5 and 15. Poor coordination and balance are often the first noticeable features. Affected individuals typically require the use of a wheelchair about 10 years after signs and symptoms appear.

About 25 percent of people with Friedreich ataxia have an atypical form in which signs and symptoms begin after age 25. Affected individuals who develop Friedreich ataxia between ages 26 and 39 are considered to have late-onset Friedreich ataxia (LOFA). When the signs and symptoms begin after age 40 the condition is called very late-onset Friedreich ataxia (VLOFA). LOFA and VLOFA usually progress more slowly than typical Friedreich ataxia.

Frequency

Friedreich ataxia is estimated to affect 1 in 40,000 people in the United States. This condition is most commonly found in people with European, Middle Eastern, South Asian or North African ancestry.

Causes

Mutations in the *FXN* gene cause Friedreich ataxia. This gene provides instructions for making a protein called frataxin. Although its role is not fully understood, frataxin is important for the normal function of mitochondria, the energy-producing centers within cells. One region of the *FXN* gene contains a segment of DNA known as a GAA trinucleotide repeat. This segment is made up of a series of three DNA building blocks (one guanine and two adenines) that appear multiple times in a row. Normally, this

segment is repeated 5 to 33 times within the *FXN* gene.

In people with Friedreich ataxia, the GAA segment is repeated 66 to more than 1,000 times. The length of the GAA trinucleotide repeat appears to be related to the age at which the symptoms of Friedreich ataxia appear, how severe they are, and how quickly they progress. People with GAA segments repeated fewer than 300 times tend to have a later appearance of symptoms (after age 25) than those with larger GAA trinucleotide repeats. The abnormally long GAA trinucleotide repeat disrupts the production of frataxin, which severely reduces the amount of this protein in cells. Certain nerve and muscle cells cannot function properly with a shortage of frataxin, leading to the characteristic signs and symptoms of Friedreich ataxia.

[Learn more about the gene associated with Friedreich ataxia](#)

- *FXN*

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- FA
- FRDA
- Friedreich spinocerebellar ataxia
- Friedrich's ataxia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Friedreich ataxia 1 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1856689/>)

Genetic and Rare Diseases Information Center

- Friedreich ataxia (<https://rarediseases.info.nih.gov/diseases/6468/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Friedreich ataxia%22](https://clinicaltrials.gov/search?cond=%22Friedreich+ataxia%22))

Catalog of Genes and Diseases from OMIM

- FRIEDREICH ATAXIA; FRDA (<https://omim.org/entry/229300>)

Scientific Articles on PubMed

- PubMed ([https://pubmed.ncbi.nlm.nih.gov/?term=\(Friedreich+Ataxia%5BMAJR%5D\)+AND+\(Friedreich+ataxia%5BTIAB%5D\)+AND+english%5BIa%5D+AND+human%5Bmh%5D](https://pubmed.ncbi.nlm.nih.gov/?term=(Friedreich+Ataxia%5BMAJR%5D)+AND+(Friedreich+ataxia%5BTIAB%5D)+AND+english%5BIa%5D+AND+human%5Bmh%5D))

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