

# [Friedreich's ataxia](https://assignbuster.com/friedreichs-ataxia/)

Friedreich's Ataxia: Treatment in Search of a Cure Ataxia refers to a wide range of ailments that are characterized by a loss of coordination in theextremities, loss of balance, and slurred or difficult speech. Ataxia, derived from the Greek word for lack of order, has many forms that exhibit similar symptoms but have different causes. Friedreich's Ataxia, named after the German neurologist that isolated and described the disease in 1863, is a rare disease that affects approximately 1 in 50, 000 people. It has its cause in a hereditary genetic disorder that must come from both parents. Commonly referred to as familial ataxia, it is caused when genetic deficiencies eliminate the production of the protein frataxin. Loss of frataxin in the system results in deregulation of iron levels in the mitochondria causing oxidative stress that kills the cells. This leads to a degeneration of the spinal cord, brain, and nerve communications to the muscles.
Symptoms are generally manifested early in life between the ages of 5 and 25. Diagnosis is usually accomplished by observing symptoms and self-reporting of symptoms. Symptoms can include spinal curvature, impaired swallowing, deformed feet, and cardiomyopathy (impaired heart muscles). In addition, with the identification of the specific genes responsible, genetic testing can be done to accurately diagnosis the disease. A family history of Friedreich's Ataxia is also an indicator used in diagnosis. As with many of the ataxias, there is not a cure but there are regimens indicated to treat the specific symptoms.
Treatment for Friedreich's ataxia generally requires the close cooperation of multiple physicians in several disciplines. The main overall goal for the patient is to remain in good health by a healthy diet and good physical exercise. Diabetes is often closely correlated with Friedreich's Ataxia and treatment consists of insulin. Further treatment is administered on an as needed basis. Propranolol, a beta blocker, may be prescribed to control muscle spasms. Beta blockers work by reducing the effect of adrenaline on the heart and lessens the 'flight or flight' syndrome and thereby reduces the tremors. Beta blockers should only be used or discontinued under the close supervision of a doctor. Amantadine, originally developed as an antiviral drug to fight influenza, has also been shown to be helpful in treating Friedreich's Ataxia. While the mechanism of action is unknown, it is believed to release brain dopamine from nerve endings making it more available to receptors. In addition, dantrolene sodium has also been used to control muscle spasms.
Other treatments involve the selective correction of the physical ailments that result from Friedreich's Ataxia. Orthopedic braces, corrective vision devices, hearing aides, and physical therapy are all used on an individual as required basis. These combinations of treatments can control the symptoms, but it can not cure the disease. There has been some hope in the field of iron chelators and antioxidants that can reduce the toxic level of iron in the mitochondria. With the recent identification of the specific genes responsible for the loss of the protein frataxia, it is hoped that the future may hold a cure. Genetic research and stem cell therapy may hold the keys to the prevention and cure of Friedreich's and many other forms of ataxia.
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