



# Friedreich's Ataxia

Friedreich's ataxia is an inherited disease that causes progressive damage to the nervous system, leading to muscle weakness, speech difficulties and heart disease.



**Q: What is Friedreich's ataxia?**

Friedreich’s ataxia is a degenerative disease caused by a defect in a gene called FXN that results in a deficiency of the protein frataxin. This deficiency damages nerve fibers in the peripheral nerves and spinal cord,

Individuals develop the disease after inheriting a defective copy of the gene from each parent. (Individuals who inherit one defective copy of the gene are carriers and can pass the mutation to their children.) Friedreich’s ataxia affects about 1 in every 50,000 people in the United States.

### **Q: What are the symptoms of Friedreich’s ataxia?**

Symptom severity and rate of progression will vary but can include scoliosis (a curving of the spine), spasticity, loss of sensation, loss of reflexes (especially in the knees and ankles), difficulty walking, fatigue, impaired coordination and balance, hearing, and vision loss, and speaking and swallowing challenges. Friedreich’s ataxia can also cause diabetes and heart disease.

Symptoms usually begin between the ages of 5 and 15 but can also appear later in life. When symptoms develop after the age of 26, the disease generally progresses more slowly.

### **Q: How is Friedreich’s ataxia diagnosed?**

Doctors will perform a physical exam to assess loss of joint sensation, absence of reflexes, balance issues, and neurological problems. Blood tests, MRI (magnetic resonance imaging) and heart tests may also be necessary. Genetic tests will confirm diagnosis.

### **Q: What is the treatment for Friedreich’s ataxia?**

While there is no cure for Friedreich’s ataxia, the FDA recently approved the first treatment for the disease: Skyclarys, an oral medication taken once-a-day, is meant to improve neurological symptoms and slow disease progression.

In general, symptoms can be managed individually. Diabetes and some associated heart problems can be treated with medication. Surgery or braces may be recommended for scoliosis and foot deformities. Physical therapy may help maintain muscle function and independence. Speech therapy can monitor and treat swallowing and speech issues. Hearing aids can be used for hearing impairment.

### **Q: Are there clinical trials for Friedreich’s ataxia?**

Yes. To find current Friedreich’s ataxia trials, visit [ClinicalTrials.gov](https://clinicaltrials.gov) (<https://clinicaltrials.gov>), a searchable website for publicly and privately supported clinical studies maintained by the National Library of Medicine at the National Institutes of Health (<https://www.nih.gov>).

Sources: National Institute of Neurological Disorders and Stroke, Johns Hopkins Medicine, MedlinePlus.

### **Need to talk to someone?**

Our Information Specialists are available to answer your questions.

Call toll-free 1-800-539-7309 Mon-Fri, 9 am-8 pm EST.

Or schedule a call or ask a question online at

<https://www.ChristopherReeve.org/Ask>.

## **Resources for Friedreich's Ataxia**

### **Friedreich's Ataxia Parents' Group (FAPG)**

<http://www.faparents.org/fapg/>

FAPG is an online support organization for parents of children with FA. The group helps limit the isolation many feel while raising children with degenerative diseases. This website provides a forum for parents to share experiences.

### **Friedreich's Ataxia Research Alliance (FARA)**

<http://www.curefa.org/>

533 W. Uwchlan Avenue  
Downingtown, PA 19335

Phone: 484-879-6160

Email: [info@cureFA.org](mailto:info@cureFA.org)

FARA features information on Friedreich's Ataxia and the related ataxias, including current research and links to related publications from scientific and medical journals. The site also offers support and information for newly diagnosed individuals, family members and caregivers.

### **Friedrich's Ataxia Research Alliance: FARA Patient Registry**

<http://www.curefa.org/patient-registry>

This international registry of individuals diagnosed with Friedreich's ataxia provides updates about current research and information about participating in clinical trials.

### **Genetic Alliance, Inc.**

<http://www.geneticalliance.org>

26400 Woodfield Rd. #189  
Damascus, MD 20872

Phone: 202-966-5557

Email: [info@geneticalliance.org](mailto:info@geneticalliance.org)

Genetic Alliance is a non-profit health advocacy organization comprised of more than 1,000 disease-specific advocacy organizations, as well as thousands of universities, private companies, government agencies, and public policy organizations. Its searchable database of diseases provides links to resources, support groups and information for patients and families.

**MedlinePlus: Friedreich's Ataxia**

<http://www.nlm.nih.gov/medlineplus/friedreichsataxia.html>

MedlinePlus, the National Library of Medicine's online resource for patients and families, offers resources with information about Friedreich's Ataxia, including symptoms, research, and clinical trials.

**Muscular Dystrophy Association (MDA)**

<https://www.mda.org/>

National Headquarters  
1016 W Jackson Blvd #1073  
Chicago, IL 60607  
Toll-free: 800-572-1717  
Email: [mda@mdausa.org](mailto:mda@mdausa.org)

MDA is a source for news and information about neuromuscular diseases (including ataxias), MDA research, and services for adults and children with neuromuscular diseases and their families. The site features a searchable database of MDA clinics and numerous publications.

**National Ataxia Foundation (NAF)**

<http://www.ataxia.org>

600 Highway 169 South, Suite 1725  
Minneapolis, MN 55426  
Phone: 763-553-0020  
Email: [naf@ataxia.org](mailto:naf@ataxia.org)

NAF, which has more than 45 affiliated chapters and support groups throughout the U.S. and Canada, works to accelerate research and improve the quality of life for people living with hereditary ataxia, a group of neurological disorders which are chronic and progressive conditions affecting coordination. Please see NAF's Ataxia Centers of Excellence at <https://www.ataxia.org/ACE/>.

**National Organizations for Rare Diseases (NORD)**

<http://www.rarediseases.org>

7 Kenosia Avenue  
Danbury, CT 06810  
Phone: 203-744-0100

NORD is a non-profit organization working to improve diagnosis, research, and treatment of rare disorders (including Friedreich's Ataxia). Individuals and families may call its Patient Information Center or submit questions online through Ask the Nurse and Ask the Genetic Counselor. A Patient Networking Program links members with other patients and families throughout the U.S.

**National Institute of Neurological Disorders and Stroke (NINDS): Friedreich's Ataxia booklet**

<https://catalog.ninds.nih.gov/publications/friedreich-ataxia>

NINDS  
P.O Box 5801

Bethesda, MS 20824

Phone: 800-352-9424

The National Institute of Neurological Disorders and Stroke is part of the National Institutes of Health. Its work focuses on advancing research and treatment of neurological disorders, including Friedreich’s ataxia.

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