Friedreich Ataxia

Friedreich 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 ataxia?

Friedreich 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 ataxia affects about 1 in every 50,000 people in the United States.

Q: What are the symptoms of Friedreich 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 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 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 ataxia?

While there is no cure for Friedreich 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 ataxia?

Yes. To find current Friedreich ataxia trials, visit 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, 7 am-8 pm EST.
Or schedule a call or ask a question online at
https://www.ChristopherReeve.org/Ask.

Resources for Friedreich 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
FARA features information on Friedreich 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 ataxia provides updates about current research and information about participating in clinical trials.

MedlinePlus: Friedreich 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 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: ResourceCenter@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
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 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.

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