



The following excerpt has been taken from the Christopher & Dana Reeve Foundation Paralysis Resource Center website.

http://www.paralysis.org/site/c.erJMJUOxFmH/b.1293667/k.CC4D/Friedreichs_Ataxia.htm

Friedreich's Ataxia

Friedreich's **ataxia** is an inherited disease that causes progressive damage to the nervous system. It can result in muscle weakness, speech difficulties or heart disease. The first symptom is usually difficulty in walking. Many persons with **ataxia** eventually use wheelchairs.

It is named after the physician Nicholas Friedreich, who first described the condition in the 1860s. Ataxia refers to coordination problems and unsteadiness, and occurs in many different diseases and conditions. In Friedreich's ataxia, there is degeneration of nerve tissue in the spinal cord and of nerves that control arm and leg movement. The spinal cord becomes thinner and nerve cells lose some of the insulation (myelin) that helps nerves conduct impulses.

Friedreich's **ataxia** affects about 1 in every 50,000 people in the United States. Males and females are affected equally. Symptoms usually begin between the ages of 5 and 15 but can appear as early as 18 months or as late as age 30.

Early signs may include foot, involuntary bending of the toes, or foot inversion (turning in). Rapid, involuntary movements of the eyeball are common. Most people with Friedreich's **ataxia** develop scoliosis (a curving of the spine to one side), which, if severe, may impair breathing.

Other symptoms include chest pain, shortness of breath, and heart palpitations. Doctors diagnose Friedreich's **ataxia** by performing a careful clinical examination, which includes a medical history and a thorough physical examination. Several tests may be performed, including electromyogram (EMG) and genetic testing.

There is currently no effective cure or treatment for Friedreich's **ataxia**. However, many of the symptoms and accompanying complications can be treated to help people maintain optimal functioning.

Friedreich's Ataxia may be inherited as an autosomal recessive trait. Cases in which a family history of the disease has not been found may indicate new genetic changes (mutations) that occur spontaneously. Friedreich's Ataxia results from mutations of a gene known as "X25, or frataxin, a protein that should normally be present in the nervous system, the heart, and the pancreas. The protein is severely reduced in people with Friedreich's **ataxia**.

Studies indicate that patients have abnormally high levels of iron in their heart tissue and that the nervous system, heart, and pancreas may be particularly susceptible to damage from free radicals (produced when the excess iron reacts with oxygen). Nerve and muscle cells also have metabolic needs that may make them particularly vulnerable to free radical damage. The discovery of the genetic mutation that causes Friedreich's **ataxia** has added new impetus to research efforts on this disease.

Sources: National Institute of Neurological Disorders and Stroke, National Organization for Rare Disorders

Websites

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

www.fortnet.org/fapg/

National Organizations for Rare Diseases (NORD) a federation of voluntary health organizations dedicated to helping people with "orphan" diseases (affects fewer than 200,000 people) and assisting the organizations that serve them. NORD is committed to the identification, treatment, and cure of rare disorders through education, advocacy, research, and service. There are more than 6,000 rare disorders, including Friedreich's Ataxia, that together affect about 25 million Americans.

www.rarediseases.org

Muscular Dystrophy Association is a primary 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 our 230 MDA clinics and numerous publications.

www.mdaua.org

National Ataxia Foundation (NAF) supports research into hereditary ataxia, a group of neurological disorders which are chronic and progressive conditions affecting coordination. NAF has more than 45 affiliated chapters and support groups throughout the U.S. and Canada. The foundation strives to improve diagnosis and develop treatment models, locating families affected by ataxia or at risk for ataxia in order to offer information and education, and identify needs and services for referral.

www.ataxia.org

2600 Fernbrook Lane
Minneapolis, MN 55447
763-553-0020

Friedreich's Ataxia Research Alliance Features information on Friedreich's Ataxia and the related ataxias, including current research, abstracts and links to publications from scientific and medical journals, ongoing studies, as well as information for researchers, patients, patient

families, and caregivers. The site also offers support and information for newly diagnosed persons.

<http://www.curefa.org/>
2001 Jefferson Davis Highway
Suite 209
Arlington, Virginia 22202
Phone: (703) 413-4468

FARA Friedrich's Ataxia Research Alliance Patient Registry. Funded by Friedrich's Ataxia Research Alliance. <http://www.curefa.org/registry/>

Genetic Alliance, Inc. is an information resource based on the notion that genetic conditions are normal and universal; that the needs of those living with genetic conditions are fundamentally the same; that genetic information is inherently personal and confidential; that families are central in the lives of those living with genetic conditions; that up-to-date information enables meaningful progress in genetic research and policy.

www.geneticalliance.org

UAB's Factsheet on Friedreich's Ataxia

<http://www.spinalcord.uab.edu/show.asp?durki=21499&site=1021&return=21497>

www.cooperative-ataxia-group.org

Cooperative Ataxia Group: Registry for North American patients

The Emory Ataxia Center

Dept. of Neurology

1365 Clifton Rd NE

Atlanta, GA 30322

Phone: 404-778-5905

Email: TheEmoryAtaxiaCenter@emory.edu

MedlinePlus: Friedreich's Ataxia

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

MedlinePlus provides resources with information about Friedreich's Ataxia. Section headings include: overview, diagnosis/symptoms, journal articles, and clinical trials.

We Move: Ataxia

www.wemove.org/ataxia

www.myspace.com/jrhink

Jason Hinckle's video My Life is posted on his MySpace account. It chronicles his life with FA.

The following books and videos are available for free loan from the PRC library. For more information, please see www.paralysis.org and click the Lending Library tab.

Books on Friedreich's Ataxia

- **Byrnes, Theresa. *The Divine Mistake: An Autobiography*.** Sydney, Australia: Pan Macmillan, 1999.
Byrnes is an Australian painter who was diagnosed with FA at the age of 17. She and her friends started the Theresa Byrnes Foundation to raise money for researching a cure.
- Danisi, Filomena. ***Battling Through Life: The Story of Filomena Danisi***. Baltimore: PublishAmerica, 2005. Autobiography.
- Lassen, Anita. ***Willpower***. Jefferson City, MO: Osage Bend Publishing Co., 2003.
Lassen is a Dane who got a college degree, married and had two daughters.
- Nance, Martha A. ***Living With Ataxia***. Wayzata, MN: National Ataxia Foundation, 1997.
- Parker, James N. and Philip M. Parker. ***The Official Parent's Sourcebook on Friedreich's Ataxia***. San Diego, CA: ICON Health Publications, 2002.
- Rieffenberger, Amanda Jean. ***Through the Eyes of a Child***. P/O Printing & Graphics, 1997.
An autobiography of a young girl. Children's book
- Schut, Henry J. ***Ten Years to Live***. Grand Rapids, MI: Baker Book House, 1978.
A true story of an entire family's battle with ataxia.
- Strode, Susan L. ***A Doe in the Wilderness***. Jefferson City, MO: Osage Bend Publishing Co., Inc., 2002.
An autobiography.

Videos

- ***Together There Is Understanding***. Minneapolis, MN
National Ataxia Foundation, 2004. Available in VHS and DVD
- ***Together There Is Hope***. Minneapolis, MN
National Ataxia Foundation, 1995

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program. You should never disregard medical advice or delay in seeking it because of something you have read in this message.