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

http://www.faparents.org/fapg/
Friedreich’s Ataxia Parents’ Group (FAPG)
FAPG 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.

http://www.rarediseases.org
National Organizations for Rare Diseases (NORD)
55 Kenosia Avenue
PO Box 1968
Danbury, CT 06813-1968
Phone: 203-744-0100
NORD is committed to the identification, treatment, and cure of rare disorders (including Friedreich’s Ataxia) through education, advocacy, research, and service. NORD offers patients information through a Patient Information Center which people can call for information. Questions can also be submitted online through Ask the Nurse and Ask the Genetic Counselor. The Patient Networking Program links members to other patients and families dealing with the same disease.

NORDS’s Rare Disease Database allows access to topics covered in the database and some associated abstracts. Full reports are available to database subscribers. Some public libraries, hospitals, and universities subscribe to NORD’s Rare Disease Database for patients and families.

http://www.mdausa.org
Muscular Dystrophy Association (MDA)
National Headquarters
222 S. Riverside Plaza, Suite 1500
Chicago, IL 60606
Toll-free: 800-572-1717
E-mail: mda@mdausa.org
MDA 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 230 MDA clinics and numerous publications.

http://www.ataxia.org
National Ataxia Foundation (NAF)
2600 Fernbrook Lane, Suite 119
Minneapolis, MN 55447
Phone: 763-553-0020
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.

http://www.curefa.org/
Friedreich’s Ataxia Research Alliance (FARA)
533 W. Uwchlan Avenue
Downingtown, PA 19335
Phone: 484-879-6160
E-mail: info@cureFA.org
FARA 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/registry/
Friedrich’s Ataxia Research Alliance: FARA Patient Registry
This registry of individuals diagnosed with Friedreich's ataxia will be used to facilitate and expedite clinical trials.

http://www.geneticalliance.org
Genetic Alliance, Inc.
4301 Connecticut Avenue, NW, Suite 404
Washington, DC 20008-2369
Phone: 202-966-5557
Email: info@geneticalliance.org
Genetic Alliance is a nonprofit health advocacy organization committed to transforming health through genetics and promoting an environment of openness centered on the health of individuals, families, and communities. The Alliance’s network includes more than 1,000 disease-specific advocacy organizations, as well as thousands of universities, private companies, government agencies, and public policy organizations. The network is a dynamic and growing open space for shared resources, creative tools, and innovative programs.

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

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**Books**


- **Byrnes, Theresa.** *The Divine Mistake: An Autobiography.* Sydney: 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.


- **Ford, Gabrielle with Sarah Thomson.** *Gabe & Izzy: Standing Up for America’s Bullied.* New York: Puffin Books, 2014. In middle school, Ford was bullied for having a degenerative muscle disease (FA). Then Ford got a dog “Izzy” who had a similar disorder. She and Izzy are on a mission to end the cycle of bullying one school at a time. For ages 8 and up.

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The following books and videos are available for free loan from the PRC library. For more information, please visit the online catalog at: [http://www1.youseemore.com/ReevePRC/default.asp](http://www1.youseemore.com/ReevePRC/default.asp)

• 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.


• Smith, Allen Graves. **Our Trials Are a Blessing...A Life Shaped by a Genetic Disease.** Orcas, WA: Chapel Valley Press, 2010. Smith was diagnosed with late-onset FA at the age of 38.


### Videos

• **Together There Is Understanding.** Minneapolis, MN. National Ataxia Foundation, 2004. VHS and DVD

• **Together There Is Hope.** Minneapolis, MN. National Ataxia Foundation, 1995

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qualified health care provider before embarking on a new treatment, diet or fitness program. You should never disregard medical advice or delay in seeking it because of something you have read in this message.

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