FARA Friedreich's Ataxia Research Alliance

curefa.org

WHAT IS FRIEDREICH'S ATAXIA?

What is FA?

Friedreich's ataxia (FA) is a genetic, progressive neuromuscular disease. People with FA experience issues with balance and coordination of movement that leads to life-altering loss of mobility. FA is life shortening and affects an estimated 5,000 individuals in the United States and 15,000 worldwide.

Symptoms of FA

All people with FA experience ataxia, loss of coordinated movement of the limbs. Sensory neuropathy — loss of sensation in the arms and legs — also contributes to the progressive loss of mobility and coordinated movement seen in FA.

Many people with FA also have heart conditions, including cardiomyopathy (thickening of the heart muscle) and sometimes cardiac arrhythmias (irregular heart rhythm).

Other symptoms of FA include
- Fatigue
- Diabetes
- Dysarthria (slurring of speech)
- Dysphagia (difficulty swallowing)
- Scoliosis
- Vision and hearing loss

FA can be diagnosed at any age, but most people are diagnosed between the ages of 5 and 15 years.

Genetics of FA

FA is caused by mutations in the **FXN** gene, which encodes the **frataxin** protein.

FA is inherited in a **recessive** pattern. Parents of someone with FA are carriers and do not have symptoms.

FXN mutations impact the mitochondria, the energy factories of cells. This leads to poor energy production and cellular damage.

There is one approved treatment for adults with FA, with many more potential treatments being researched for children and adults.

The Friedreich's Ataxia Research Alliance (FARA) is a national, public, 501(c)(3), non-profit, tax-exempt organization dedicated to the pursuit of scientific research leading to treatments and a cure for Friedreich's ataxia. For more information on FA, visit curefa.org or email info@curefa.org.

ABOUT FARA

Who is FARA?

The Friedreich's Ataxia Research Alliance (FARA) is a national, public, 501(c)(3), non-profit organization dedicated to the pursuit of scientific research leading to treatments and a cure for Friedreich's ataxia.

FARA's Mission is to marshal and focus the resources and relationships needed to cure FA by raising funds for research, promoting public awareness, and aligning scientists, patients, clinicians, government agencies, pharmaceutical companies and other organizations dedicated to curing FA and related diseases.

FARA's leadership framed the organization's strategic priorities and activities into the following four essential pillars.

- **Attracting & Facilitating Collaboration in the Community:** FARA assembles all stakeholders researchers, physicians, patient families, industry and government partners – to share insights, new research and build partnerships that unite us as one well-aligned and stronger FA community.
- **Deploying Financial Resources:** FARA deploys financial resources through a competitive, peer reviewed research grant program as well as FARA directed research.
- **Creating Domain Resources:** FARA builds resources that help advance all therapies in the field. Examples of domain resources include the FA Global Patient Registry, the FA-Integrated Clinical Database, and bio and cell repositories.
- **Sharing Knowledge & Know-How:** FARA believes that sharing the knowledge from our grant program, research conferences, academic and industry partners, and patient families optimizes our pace and enables the efficient use of resources.

These pillars provide the tools, the knowledge and the collaborative relationships that advance therapeutic candidates in the drug development pipeline – to slow, stop, reverse, and cure FA.

How to support FARA

Your support allows us to sustain and increase our investment in these critical areas of work today, so we may ensure a future for our patient families without the devastation of FA. Thank you for being an active partner in our effort to treat and cure Friedreich's ataxia. If you would like to make a gift to FARA, please visit curefa.org/donate.