Biogen

WE ARE FA

Your Guide to Friedreich Ataxia: Understanding Symptoms, the Path to Diagnosis, Monitoring Progression, and Building Your Care Team

WE ARE FA

Friedreich Ataxia at a Glance

Friedreich ataxia (FA) is a rare, life-shortening genetic condition that progressively damages the nervous system and causes loss of muscle control over time.

While each person's experience with FA is different, understanding the core characteristics of this disease can help you make informed decisions about your healthcare.

FA is rare:

FA is considered a rare disease, affecting about 1 in 50,000 people. More than 5000 people in the United States and 15,000 worldwide may have FA. It is the most common inherited form of ataxia.

FA is genetic:

The condition stems from specific changes in the frataxin (*FXN*) gene. For FA to develop, a person must inherit altered copies of this gene from both parents. FA is a recessive condition, meaning it can be passed down through families even without an obvious family history, so siblings and other family members should consider genetic counseling and testing.

FA is progressive:

Symptoms usually first appear between the ages of 10 and 15. The disease progressively damages the nervous system, leading to loss of muscle control and ambulation. Eventually, FA symptoms may significantly impact the ability to perform everyday activities as physical abilities become more impaired.

Learn more about FA signs, symptoms, and genetic testing at WeAreFA.com

The Genetics of FA

The genetic mutation that causes FA occurs in the *FXN* gene and involves a DNA sequence called the GAA triplet-repeat expansion. In people without FA, this GAA sequence repeats 33 or fewer times. In people with FA, it can repeat hundreds or even more than a thousand times.

The number of GAA repeats is closely linked to how old you are when symptoms start and how quickly FA might progress. A higher number of repeats typically means earlier onset and faster progression, while a lower number of repeats usually means later onset and slower progression.

The *FXN* mutation causes a waterfall of effects that lead to the symptoms of FA

1. The *FXN* mutation results in reduced production of a protein called frataxin.
2. Frataxin deficiency can lead to too much iron in the mitochondria, reduced energy production, and oxidative stress.
3. A protein called Nrf2 has also been shown to be deficient in FA.
4. When Nrf2 doesn't work properly, it may contribute to the worsening effects of oxidative stress inside cells such as nerve cells.
5. Eventually, too much iron and oxidative stress can cause nerve cells to die and may lead to symptoms like loss of muscle control.

Signs and Symptoms of FA

FA affects people differently, but while everyone's experience will be unique, there are some common things that doctors may look for when considering FA.

The first signs of FA typically appear between the ages of 10 and 15, but symptoms can emerge at almost any age. People with FA often first see a doctor because they feel overly tired and have trouble with their balance or walking and aren't sure why. The **classic early symptoms of FA** can include:

- Falls with gait ataxia
- Lack of balance (poor proprioception)
- Loss of sensation (neuropathy)
- Loss of reflexes (areflexia)
- Chronic fatigue

Diagnosing FA

Diagnosing FA can be challenging and may take 3 years on average. Once you've decided to seek medical help, there are specific tests the doctor may perform that can help reach a diagnosis:

Reflexes in the lower legs: This test involves tapping your knee with a reflex hammer to see how your leg reacts.

Vibrational sense in the feet: In this test, a special tuning fork is struck and placed on your foot to see if there might be nerve damage in your lower limbs.

Clarity of speech: Some people with FA slur their words. While not a universal symptom, your physician will likely check for speech difficulties.

Thickness of the spinal column: An MRI can show whether certain parts of the spinal column appear narrower than usual, which can be a sign of FA.

Choosing the right genetic test matters

Not all genetic tests are designed to diagnose FA. In particular, tests called whole exome or next generation sequencing panels may not accurately detect FA. To confirm FA, a specialized test called the **GAA repeat expansion test** is needed. Even if you've already had a genetic test for ataxia, you might still need this specific test to accurately diagnose FA.

Is it FA?

Misdiagnosis or delayed diagnosis is common, especially among older patients or those with nonspecific symptoms. FA can mirror symptoms of other neurological conditions like multiple sclerosis (MS) in the early stages of the disease. FA may also be misdiagnosed as one of several types of ataxia.

Ask your doctor about FA Identified, a no-charge genetic testing program sponsored by Biogen and offered through PreventionGenetics. If your doctor isn't familiar with the program, they can learn more at FAIdentified.com.

FA IDENTIFIED

FA Progression

Everyone with FA will see their symptoms progress over time. Some may progress more quickly or slowly than others, and not everyone will experience the same symptoms. Your doctor may perform certain tests to monitor your progression because FA symptoms can significantly impact your ability to perform everyday activities.

FA symptoms become apparent:
falls, clumsiness, feeling unbalanced, loss of sensation, tiredness

Hands and arms
become less coordinated, affecting the ability to get dressed or brush teeth

Lower limb coordination
continues to decline

Speech
starts to sound slurred

Early diagnosis can help you better manage life with FA. Talk to your doctor about your symptoms and how you can be proactive in your treatment journey

FA and mobility

As people with FA become less coordinated, they may start using more assistive devices to help them get around and perform their daily activities. For example, you may progress from using a cane to different mobility aids such as: walker, rollator (wheeled walker), if we can and wheelchair.

On average, people with FA will need to use a wheelchair about 10 to 15 years after their first symptoms show up. Later in the course of disease, people with FA can become incapacitated and unable to get around on their own or perform many activities of daily living.

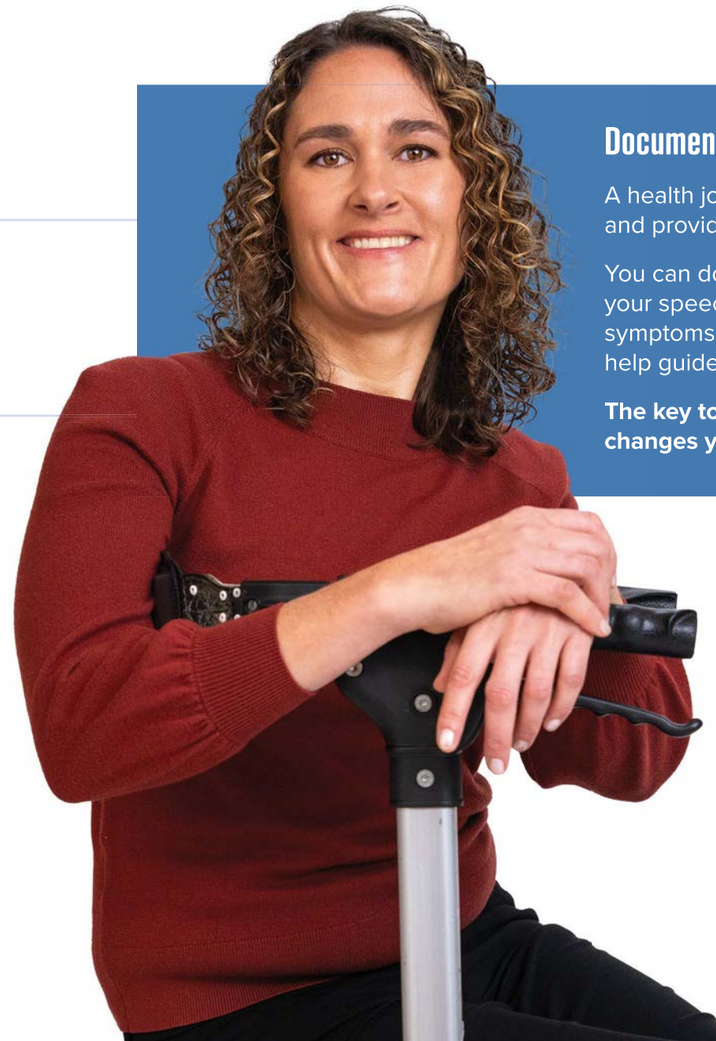
FA and Other Conditions

Certain other conditions are also more common among people with FA.

Diabetes affects many people with FA—up to around 30%, according to some estimates.

Diagnosis of **scoliosis** often coincides with the diagnosis of FA.

Cardiomyopathy is a thickening of the walls in the heart that makes it more difficult for the heart to pump blood over time and can lead to premature death. Nearly 70% of patients with FA in a large study were found to have evidence of cardiomyopathy.



Document your FA experience

A health journal can help you monitor changes in your abilities and provide valuable information to share with your doctor.

You can document how you're feeling, your energy levels, how clear your speech is, anything to help your doctor understand how your symptoms have changed since your last visit. Your observations can help guide the development of a personalized care plan.

The key to a good health journal is consistently noting changes you experience.

Building a Healthcare Team

A team as unique as you

Because FA affects many different parts of the body, you may want to put together a team of healthcare specialists, each working together to address different needs that may arise during the FA journey.

Primary Care Provider

The primary care provider coordinates overall health management and helps maintain continuity of care across all specialists. They manage routine health needs, help navigate referral processes, and ensure medications work safely together.

Neurologist

The neurologist specializes in monitoring FA's effects on the nervous system. They track disease progression, assess neurological changes, and work closely with other specialists to adjust treatment plans based on individual symptoms and challenges.

Cardiologist

The cardiologist focuses on protecting and monitoring heart health, which is essential in FA management.

Physical Therapist

Physical therapists play a crucial role in maintaining strength, balance, and mobility.

Occupational Therapist

Occupational therapists help maintain independence by recommending practical modifications and equipment to make everyday activities more manageable.

Speech Therapist

Speech therapists address both communication and swallowing challenges that may develop with FA.

Mental Health Professional

Mental health professionals support emotional well-being throughout the FA journey for individuals living with the condition and for their families.

Genetic Counselor

Genetic counselors help families understand FA's inheritance pattern and implications for family planning.

While not everyone needs every specialist at once, understanding each potential team member's role can help you figure out what kind of care you need and when

Connecting With the FA Community

The FA community can guide you to others who understand your health journey and can offer insights into life with FA.

National groups offer comprehensive support, education, and research initiatives across all ataxias, with helpful educational content through podcasts, books, and other media that reflect real experiences with FA.

FARA is a national nonprofit organization dedicated to the pursuit of scientific research leading to treatments and a cure for FA.

NAF is an organization working to improve the lives of people with ataxia by funding research, supporting families, and collaborating with partners to accelerate treatment development.

MDA is the #1 US voluntary health organization for people living with neuromuscular diseases. For 75 years, MDA has led the way in accelerating research, advancing care, and advocating for families.

> There's a whole community of people with FA out there who are ready to share their experiences and offer advice on how to navigate the ups and downs of FA

Connect through clinical research

Clinical trials play a vital role in exploring new therapeutic approaches to treat FA, measuring disease progression, and examining how FA affects different individuals. Participation in clinical trials is a personal choice, but it's a choice that can contribute to advancing FA research and treatment options.

For information about current trials, talk with your healthcare team or connect with FA advocacy organizations like FARA. You can also visit ClinicalTrials.gov and search for "Friedreich ataxia" to see all the current and ongoing trials for FA.

Talk to your doctor today to learn more about genetic testing, disease progression, and your FA journey.

Visit
WeAreFA.com
for more information

WE ARE FA

225 Binney Street, Cambridge, MA 02142
FA-US-0232v2 06/25

Biogen