Where to Begin?

A Guide for Individuals Recently Diagnosed with Friedreich's Ataxia

A diagnosis of Friedreich's ataxia (FA) ushers in a wide array of emotions and life changes that impact each individual and family in unique ways. This guide can help you navigate through this complex time by providing a starting point for learning about FA. Through this guide, you can also find ways to connect with the vibrant and engaged FA community, which includes individuals and families affected by FA, physicians, researchers, and others who are working together to find treatments for FA.

Although FA is a rare disease, there are about 15,000 people with FA in the world, with 5,000 people living in the United States. Many members of the FA community have stood exactly where you stand now, facing a new diagnosis and the ocean of information and emotions that accompany it. We hope you will be able to lean on the strong network of support and friendship you can find in the FA community as you begin your journey with FA.

This guide does not represent an exhaustive source of information on FA. If you have specific questions, want to learn more, or wish to connect with others diagnosed with FA, visit curefa.org or reach out to the Friedreich's Ataxia Research Alliance (FARA) at info@curefa.org or 484-879-6160.

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FARA Friedreich's Ataxia Research Alliance

Symptoms of FA

Neurological Symptoms

Early symptoms of FA include **trouble walking** and **frequent tripping**. People may also have **difficulty writing** and **reaching for or grasping objects.**

Some symptoms stem from changes in the nervous system, including:

- ***Ataxia*** – loss of coordinated movement of the limbs
- ***Reflex and sensory loss*** – difficulty sensing vibrations and position of the limbs

FA is a progressive disease, which means symptoms change or worsen over time. As time passes, someone with FA will likely transition through different phases of mobility such as:

- Using walls, furniture, or loved ones as support while walking
- Using a cane or a walker
- Using a wheelchair for longer distances
- Using a wheelchair full time

Most people with FA experience ataxia as their first symptom. Ataxia is the loss of coordinated movement of the limbs. Although it seems simple, voluntary movement, such as taking a step or reaching out to grasp a cup, relies on several types of information that the brain must properly piece together. When reaching for a cup, for example, the reaching arm and the brain communicate to figure out where the arm is in space compared to the cup, the brain tells the arm to move, and the arm confirms its movement with the brain.

Ataxia is caused when certain areas of the nervous system are not functioning well. This prevents the brain from properly sending messages back and forth to the limbs. In FA, specific regions of the nervous system involved in communicating these messages, the cerebellum (in the brain) and the dorsal root ganglia (near the spinal cord), are especially affected by the genetic mutation that causes FA (see the Genetics of FA section). This disrupted communication between the brain and the limbs leads to symptoms of poor coordination and balance.

People with ataxia rely on their vision and touch senses to figure out where their body is in space and to compensate for the jumbled communication between the brain and limbs. You might notice

yourself touching or grasping furniture, walls, or loved ones to support yourself when walking. This is not because your muscles are weak, but because you are using these objects as a reference point to help your brain figure out where your body is in space. Similarly, you may notice that you're clumsier in low light or when you close your eyes. This is because you are relying on visual cues to help your brain figure out where you are. Using nightlights or flashlights in the dark or trying to keep your eyes open in the shower might help to prevent falls.

FA is a progressive condition which means that the symptoms change or worsen over time. Each person living with FA has their own unique experience of the disease, and not all individuals will experience every symptom or the same rate of progression. On average, the neurological symptoms of FA progress to the point of needing a wheelchair for mobility within ten years of diagnosis.

Heart Conditions in FA

Many people with FA have ***cardiomyopathy***, or the thickening of the heart muscle. This heart condition is diagnosed through an echocardiogram (ultrasound of the heart). Cardiomyopathy can make it difficult for the heart to pump blood efficiently and can sometimes progress to heart failure over time. People with FA who have cardiomyopathy frequently don't experience heart-related symptoms. Those who do have symptoms related to cardiomyopathy may experience chest pain, shortness of breath, and dizziness.

Some people with FA also have ***cardiac arrhythmias***, or an irregular heartbeat. This can include a heart that has episodes of beating too slowly or quickly, or a heart that sometimes beats in a chaotic rhythm. Arrhythmias are diagnosed through an electrocardiogram (ECG/EKG), a test that measures the heart's electrical activity. Someone with FA who has cardiac arrhythmia may experience chest pain, shortness of breath, or heart palpitations (feeling the heart "fluttering" or "skipping a beat").

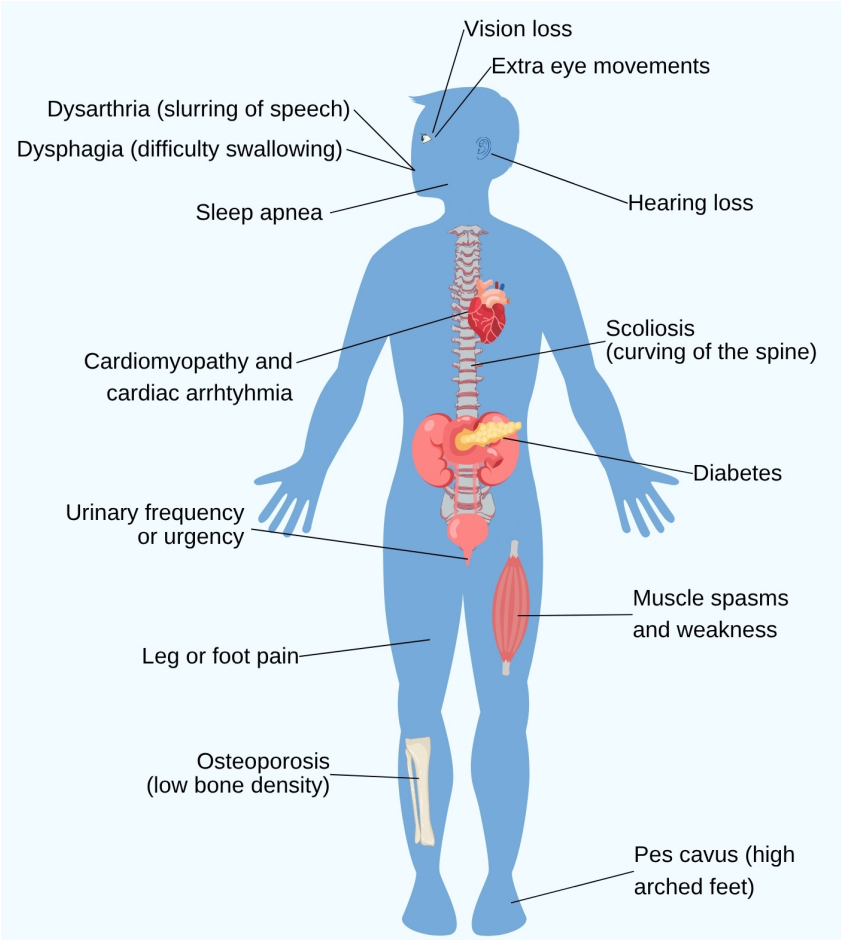
Many people with FA who get an EKG test will have an abnormal result that does not always correlate to a diagnosis of cardiomyopathy or cardiac arrhythmia. This type of abnormal result does not require any type of clinical management or medication.

An FA diagnosis is not a contract on the life you'll experience. Like any other adversity, it will definitely change some details, but ultimately you are in charge of creating the outline of your life.

–Shandra
Individual with FA

Other Symptoms

FA is a multi-system condition which means that it can affect multiple organs. This leads to different types of symptoms occurring over time. The diagram to the right shows different symptoms of FA. It's important to remember that not every person with FA will experience every symptom. Symptoms may appear over time, not necessarily all at once. There is no predetermined course of symptom development or progression.



Many people with FA also experience fatigue, depression and anxiety.

Dysphagia, or difficulty swallowing, may present as choking while drinking liquids.

Diabetes is caused when the body does not regulate blood sugar levels properly. In FA, diabetes can present in childhood or adulthood and can be managed with standard treatment approaches.

Fatigue impacts many people with FA. More than just general tiredness, fatigue is a type of exhaustion that can prevent someone from carrying out everyday activities. The cause of fatigue in FA is not fully understood but is likely due to a combination of factors. This includes both the large amount of work the body must invest to compensate for balance issues, as well as the fact that the genetic mutation that causes FA directly impacts cellular energy production (see Genetics of FA).

Depression and anxiety are not uncommon for people with FA. It is unclear if mental health issues are a primary symptom of FA caused by the impact of the genetic mutation, or if they are a secondary symptom stemming from the difficulties of living with a complex, rare disease.

Scoliosis occurs in about 70% of people with FA. Sometimes scoliosis may get worse over time and require corrective surgery.

Vision and hearing loss usually are not present at diagnosis but can develop over time.

Late Onset FA

About 25% of people with FA first develop symptoms in adulthood. This is referred to as late onset of FA. Individuals with late onset FA typically experience slower progression of neurological symptoms and usually are not affected by cardiomyopathy.

Example Individuals with FA

FA affects everybody differently. Not every person will develop every symptom, and not every person will experience the same pattern of progression. For example, these three individuals with FA are all affected differently by the disease:

Grace was diagnosed with FA at age 5. She is now a 21-year-old college student who loves to read. FA has had a significant impact on her physically. She has had surgery for scoliosis, uses a wheelchair for mobility, and needs assistance with daily activities.

Erin developed symptoms of FA in her early 20s and was diagnosed at age 38. She is now 45 and she enjoys doing arts and crafts and spending time with her husband, son, and extended family. She uses a mobility scooter to conserve energy. She has mild scoliosis and experiences muscle cramps in her arms, legs, and feet. She does not have cardiomyopathy.

Byron, Keith and Stuart Andrus;
photo courtesy of Raychel Bartek

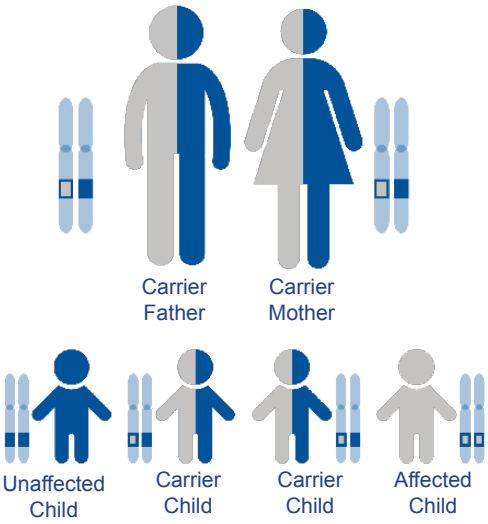
Keith was diagnosed with FA at age 11. He loved music, collecting sneakers and getting tattoos. He experienced all the symptoms of FA: scoliosis, diabetes, and vision and hearing loss. He passed away at 24 due to complications from cardiomyopathy.

Genetics of FA

FA is a genetic, neurological disorder caused by changes in the FXN gene, which makes a protein called frataxin. Frataxin is used by mitochondria, the energy generating components of cells. FA is caused by a specific type of genetic change known as a GAA repeat expansion and is inherited in an autosomal recessive pattern. Continue reading below to learn about GAA repeat expansions, autosomal recessive inheritance, and how mitochondria use frataxin to provide cells with energy.

Genes & Autosomal Recessive Inheritance

Our DNA, or genetic code, is organized into units called genes. Genes contain the instructions for making protein, the building block of cells and tissues. Humans have two copies of every gene. One is passed down from the biologic mother, and the other is passed down from the biological father. FA is an autosomal recessive condition, which means it occurs when non-functioning copies of the FXN gene are passed down from both parents. A person with FA has two non-functioning copies of the FXN gene.

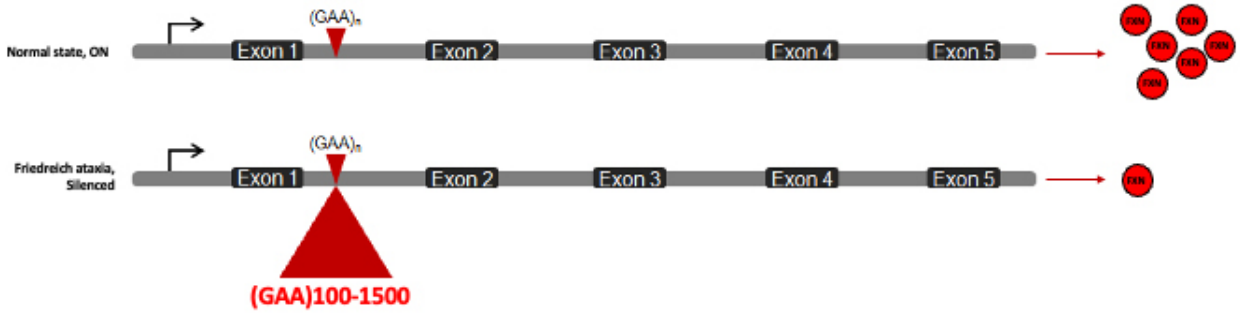


Parents of a person with FA are called carriers: they each have one working copy and one non-functional copy of the FXN gene. In the United States, it is estimated that 1 in every 100 people, or 1% of people, are carriers of FA. When both parents are carriers, each of their children has a 1 in 4, or 25%, chance of having FA.

Because FA is an inherited disease, there may be other people in your family who should consider genetic testing, including your siblings. Genetic testing is a personal choice that requires careful consideration and discussion. Visiting with a genetic counselor may help you and your family make these important decisions.

GAA Repeat Expansion

All genes are composed of a genetic alphabet containing four letters: C, G, A, and T. The spelling of the FXN gene includes a repetition of the three letters “GAA.”

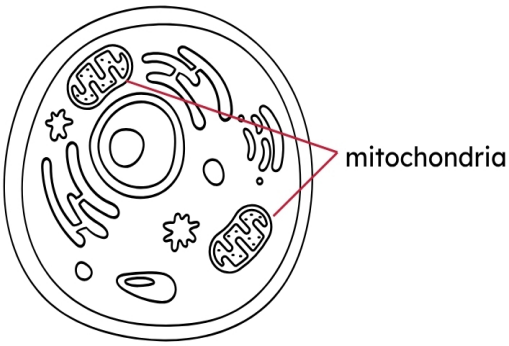


People unaffected by FA usually have between seven and 30 GAA repeats in the FXN gene. FA is caused when this stretch of GAAs is expanded. Symptoms of FA become noticeable in individuals with 66 or more GAA repetitions on both copies of the FXN gene. Higher numbers of repetitions have been linked to earlier age of onset and increased symptoms of FA.

Most people with FA have two FXN genes with over 100 repeats. But FA can also be caused by other types of changes in the FXN gene including point mutations and deletions. In these cases, the correct letter of the DNA sequence is replaced by a different letter or letters are deleted from the gene. Four to five percent of people with FA have one FXN gene with over 100 repeats and one FXN gene with another type of mutation.

Frataxin & the Mitochondria

The FXN gene encodes the instructions to make a protein called frataxin, which supports the function of a part of the cell called the mitochondria. The mitochondria create energy for the cell to use in the form of a molecule called ATP. In the mitochondria, frataxin helps to transport iron molecules and form iron-sulfur clusters. These iron-sulfur clusters play a vital role in energy production within the mitochondria. When this system is working well, enough ATP energy is made to keep cells functioning normally.



Human Cell

> Don't put your life on hold until they find a cure. Learn to live the best you can in the reality we have now.
>
> –Katherine
> *Individual with FA*

The genetic mutations that cause FA lead to a deficiency of frataxin, which prevents the mitochondria from producing enough ATP to keep the cell functioning well. Low levels of frataxin also lead to a buildup of excess iron within the cell. Too much iron can trigger the formation of reactive oxygen species, toxic molecules which damage the cell and lead to cell death. Because of its relationship to energy production and cell health, low levels of frataxin especially impact high energy organs such as the nervous system and the heart. In FA, cells in the brain, spinal cord, and heart muscle do not have enough energy and are damaged by reactive oxygen species. Over time, this damage and lack of cellular energy causes the symptoms of FA.

Management and Treatment of FA

FA Care Team

Most people with FA receive care from a variety of specialists to manage and monitor their symptoms. After a new diagnosis, the list of specialists to see can be overwhelming and learning how to best manage all of these visits can take time and patience.

Managing FA

Here are the specialists to see after a new diagnosis:

- ***Neurologists*** are typically the primary providers for people with FA because they are able to track symptoms related to ataxia and reflex and sensory loss. Upon diagnosis, your neurologist should perform a neurological assessment to understand your current symptoms and functional level. As your main FA care provider, your neurologists will likely be the doctor referring you to the other specialists who will be part of the team. Neurologists with FA experience can be found through the FA Global Clinical Consortium.

- ***Cardiologists*** work with individuals who have FA to monitor the heart for signs of cardiomyopathy (thickening of the heart muscle) or arrhythmias (irregular heartbeats). After diagnosis a cardiologist should test your heart function with an echocardiogram (an ultrasound of the heart) and an EKG (a measurement of the heart's electrical activity). For some individuals, cardiologists manage symptoms of cardiac disease with medications and other strategies.

- ***Physical and occupational therapists*** work with people with FA to build strength and learn how to modify different daily tasks that are impacted by symptoms, so it is recommended to be evaluated by these specialists after diagnosis.

- ***Primary care physicians***, such as your family doctor, are important members of your care team. It's important to have a local doctor who knows you well and can help with urgent situations or run-of-the-mill illnesses and injuries.

In addition to the exams done by a neurologist and cardiologist, it is important to have screening for scoliosis and blood glucose assessed after an FA diagnosis. Scoliosis and blood glucose screenings can usually be ordered or performed by your neurologist or primary care doctor. Your FA specialist may recommend other exams, specialists, or procedures that are unique to you. **After these initial evaluations, neurological assessment, heart function tests, physical and occupational therapy assessments, scoliosis assessment, and blood glucose evaluation should be repeated every year.**

Each person with FA has their own unique set of symptoms. These additional evaluations may be needed from other specialists based on an individual's symptoms and disease progression.

- ***Speech therapists or speech language pathologists*** can do assessments of speech and swallowing if you are experiencing dysphagia (difficulty swallowing) or dysarthria (slurred speech). They would also help to treat these symptoms.
- ***Orthopedists*** can help manage scoliosis (curving of the spine) or pes cavus (high arched feet).
- ***Endocrinologists*** can help manage diabetes, growth differences, and low bone density (osteoporosis).
- ***Mental health professionals*** like therapists, psychologists, and psychiatrists, can help you manage mental health issues.
- ***Audiologists*** can assess for hearing loss and suggest adaptations.
- ***Ophthalmologists*** can assess for vision loss and suggest adaptations.
- ***Geneticists and genetic counselors*** can help you and your family understand how FA is inherited and facilitate genetic testing for other family members.
- ***Pulmonologists and sleep specialists*** can help assess for and treat sleep apnea.

Lastly, the most important member of the FA care team is you. Your input and advocacy are integral to managing FA. Some of your new specialists may not be familiar with FA, and it will fall to you to introduce them to different aspects of the condition. Don't be afraid to speak up if you feel like a care plan isn't going in the right direction; you are the expert in the room when it comes to you or your FA symptoms and experiences. As you work to build your FA care team, you may need to try different doctors until you find those who are the right fit for your team.

If you are meeting with a new clinician who is unfamiliar with FA, it may be helpful to refer them to resources that explain the condition including this guide and the FA Clinical Care Management Guidelines, which are intended to help qualified healthcare professionals make informed care plans for individuals with FA.

SKYCLARYS® (Omaveloxolone)

As of 2025, SKYCLARYS is approved for people living with FA in the U.S., E.U., Canada, UK, and Brazil who are ages 16 and older. While approval is an important milestone, individual countries will still determine access through their own processes. Biogen, the company that owns and manufactures the drug, is currently undertaking efforts to expand approval in additional countries and in children, and is working to address gaps in access. You can learn more about SKYCLARYS on FARA's website.

Approved Treatment

SKYCLARYS improves mitochondrial function through several mechanisms, including increasing production of antioxidants, molecules that protect the mitochondria from reactive oxygen species. This allows mitochondria to function better even with deficient frataxin levels. Clinical trials showed that SKYCLARYS improves and slows progression of FA symptoms.

Future Treatments: Research and Clinical Trials

SKYCLARYS is not a cure for FA, but an important first step in treatment. Physicians and scientists believe multiple treatments will be needed to address all aspects of FA and stop the progression of the disease.

Participate in Research

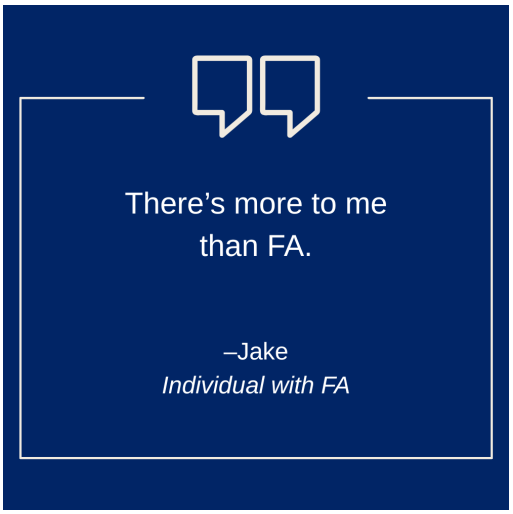
The Friedreich's Ataxia Research Alliance (FARA) is dedicated to advancing treatments for FA. Currently, there are several potential treatments being researched and pursued. Visit FARA's website to learn more about the drug development pipeline.

The FA community's participation in research is integral to advancing the development of future treatments. Two types of clinical research people with FA can participate in are interventional clinical trials and observational clinical studies. **Interventional clinical trials** involve testing a treatment in patients to collect data about the safety of the treatment and how well it works. **Observational clinical studies** do not involve testing a treatment but rather focus on collecting data about the natural history of FA (symptoms and progression) and potential biomarkers. The observational natural history study for FA, UNIFAI, is open to all individuals with FA to join. Our current understanding of FA, the approval of Skyclarys, and the approval of future treatments are owed to all of the people with FA who have participated in clinical studies and trials. Learn more about participating in FA research on FARA's website.

Exercise, Nutrition, and Supplements

Exercise and Physical Therapy

Exercising regularly is an important part of maintaining health for everyone. Because FA affects balance and coordination, exercises focused on improving mobility, balance, and strength are particularly useful for someone with FA. Exercise that strengthens muscles may help you compensate for balance and coordination issues, and exercise may also help improve fatigue and sleep. Exercises that are recommended for people with FA include strength training of the arms, legs and trunk (core), cardiovascular exercise (such as swimming, walking, or biking), balance exercises, and stretching.



Adaptive exercise equipment, including recumbent trikes and hand cycles, are great options for individuals who use mobility aids. Check out the Ataxian Athlete Initiative, which helps people with ataxia access adaptive cycling equipment.

Physical therapists are integral members of the FA care team who can teach you exercises and develop an exercise plan tailored to your specific needs and goals. Regular physical therapy sessions can help people with FA maintain strength. Physical therapists can also help address any new mobility issues that may arise.

Nutrition

Nutrition is another aspect of health that impacts all people. Eating a balanced diet may help to increase energy levels, aid digestion, and improve mood. Like all other aspects of FA, nutritional needs are unique, and no two people with FA will thrive on the same exact diet. Although balanced diets are usually considered high in protein and fiber and low in simple carbohydrates, it is important to work with your care team to find the diet best for you.

Additionally, people with FA may experience dysphagia, or trouble swallowing. This symptom often presents as choking when drinking water or other thin liquids. Using straws or liquid thickeners may help avoid choking.

Supplements

Supplements are vitamins and minerals that our bodies need to properly function and stay healthy. Most of us get enough of these vitamins and minerals from our regular diets. If someone is not getting enough of a specific vitamin or mineral from their diet, they may need to add a "supplement" of the lacking nutrient.

Some individuals with FA take the antioxidant supplements idebenone, CoQ10, or vitamin E. Antioxidants are thought to support mitochondrial health by protecting against damaging free radical molecules. Because the root cause of FA is mitochondrial dysfunction, several research studies have investigated the use of these supplements as treatments for FA. Studies in animal and cell models suggested the supplements may benefit people with FA. Clinical trials where these supplements were tested in people with FA showed that the supplements were safe to consume. However, the clinical trials were not able to prove that these supplements slowed the progression of FA symptoms. The clinical trials were small and short in duration. Because of the small number of trial participants and the fact that FA symptoms tend to progress slowly, it is possible that these supplements do have a positive impact that the trials were simply unable to capture.

Because the trials could not prove that these supplements slow progression of FA symptoms, idebenone, CoQ10, and vitamin E are not recommended or approved treatments for FA. However, because they are safe to consume, theoretically linked to mitochondrial health, and can be acquired without a prescription from a doctor (over the counter), some individuals with FA take these supplements.

Even though you usually don't need a prescription to get these supplements, it is important to speak with your doctor or care team before taking any supplements or vitamins. Your doctor can help determine if these supplements may benefit you and ensure that the supplements will not interact with any other medications or vitamins you may be taking.

The FA Community: You Are Not Alone

After a new diagnosis, it is common to feel isolated and struggle to find support. Despite the fact that FA is classified as a rare disease, there is a strong FAmily of other people with FA, their family members and friends, and others ready to support you. It can be overwhelming to jump right into a community you were never expecting to be a part of, but these resources are here for you whenever the time is right.

Contact FARA at info@curefa.org or 484-879-6160 for support.

Connecting With the FA Community

FA Parent's Group: For parents or caregivers of children with FA or other pediatric onset ataxias

FARA's FA Ambassador Program: For individuals with FA 16 and older who want to volunteer to support FARA's mission to cure FA

FA Hangouts: social video calls for adults with FA hosted by FARA ambassadors

Teen Hangouts: a safe space to hangout online for teens with FA, moderated by two FARA ambassadors

Connect

The National Ataxia Foundation has several support groups for children and adults affected by ataxia as well as family members and caregivers.

The FA App helps to connect and empower individuals and families with FA worldwide, and Facebook has several FA support groups, including international groups.

FARA Community Events including fundraisers and educational receptions are a great way to meet others with FA in person.

Community Stories

Meet the Community on FARA's website

Videos from the FA Community on FARA's YouTube page

Two Disabled Dudes: podcast hosts Kyle and Sean, who have FA, discuss living life beyond circumstances and have conversations with other members of the disability and rare disease communities

International Ataxia Organizations

Argentina: La Asociación Civil de ATaxias de ARgentina

Australia: Fara Australia

Brazil: Ataxia de Friedreich Brasil

Canada: Ataxie/Ataxia Canada

Czechia: FRIEDA

Denmark: FORENINGEN for ATAKSI HSP

Global FA Patient Organizations

Europe: Euro Ataxia

France: Association Francaise de l'Ataxie de Friedreich

Germany: Friedreich Ataxie Fordeverein e.V. ; Deutsche Heredo-Ataxie Gesellschaft

Greece: Hellenic Friedreich's Ataxia Association

India: Ataxia Awareness Society

Ireland: FARA Ireland

Italy: Italian Association for the Fight Against Ataxia Syndromes (AISA)

Lebanon: Friedreich's Ataxia Lebanon Association

New Zealand: FARA New Zealand

Portugal: Associacao Portugesa de Ataxias Hereditarias

Spain: La Federacion de Ataxias de España

Sweden: Bota FA

United Kingdom: Ataxia U.K.

Ways to Get Involved

- Consider participating in research to advance our understanding of FA and potential treatments.
- Get involved in FARA's fundraising efforts by participating in or volunteering at a fundraising event or donating online.
- Advocate with FARA to support research and healthcare access for FA and other rare diseases.
- Attend a FARA Research Reception or FA community event.

What is FARA?

The Friedreich's Ataxia Research Alliance (FARA) is a nonprofit 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, individuals and families with FA, clinicians, government agencies, pharmaceutical companies and other organizations dedicated to curing FA and related diseases.

What You Can Do Next

- Start building your FA care team by reaching out to specialists and planning appointments
- Download the Clinical Care Guidelines to give to doctors that may not be as familiar with FA
- Consider joining the UNIFAI natural history study to contribute to our understanding of FA
- Connect with others in the FA Community through FA Hangouts or our Meet the Community series
- Attend a FARA event such as a symposium, rideATAXIA, or other local event
- Sign up for FARA's newsletter to stay up to date on current research, trials, and events