WE ARE FA

Patients pictured are paid spokespersons for Biogen.

Genetic Testing for FA:
What You Need to Know

Friedreich ataxia (FA) is a rare, life-shortening genetic condition that progressively damages the nervous system and causes loss of muscle control over time. Genetic testing may help provide important insights into FA and other genetic ataxias.

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FA and Genetic Testing

Understanding ataxia

Ataxia symptoms—such as difficulty walking, clumsiness, lack of fine motor coordination, and slurred speech—may be caused by an underlying genetic condition.

There are 3 types of ataxias:

- **Acquired ataxia** develops due to outside factors like a stroke, tumor, or vitamin deficiency
- **Hereditary ataxia** is passed down through families through genetic mutations
- **Sporadic ataxia** appears without evidence of being passed down through family

FA is the most common form of hereditary ataxia, affecting more than 5,000 patients in the United States. Genetic testing can help determine whether FA may be the cause of your ataxia symptoms even if you don't have a family history.

FA is caused by a mutation in the frataxin (*FXN*) gene

The mutation in the *FXN* gene that causes FA is called a GAA triplet-repeat expansion. Usually, the GAA triplet repeats 33 or fewer times. In people with FA, the GAA triplet can repeat hundreds or sometimes more than a thousand times.

The *FXN* mutation results in reduced production of a protein called frataxin. Frataxin deficiency can lead to too much iron in the mitochondria, reduced energy production, and oxidative stress. Eventually, too much iron and oxidative stress can cause nerve cells to die and may lead to symptoms like loss of muscle control and continued disease progression.

FA is a progressive condition, meaning symptoms will worsen over time. Eventually, FA symptoms may significantly impact your ability to perform everyday activities as physical abilities become more impaired.

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.

Get Answers With FA Identified

What is FA Identified?

FA Identified is 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**.

The FA Identified test includes:

- GAA triplet-repeat expansion test—the key diagnostic tool for FA
- Testing for other *FXN* mutations—included if additional testing is needed

Who qualifies?

- Individuals aged 16 and older who have or are suspected of having FA
- Must be in the United States or Puerto Rico and ordered by a qualified healthcare provider

Steps to getting tested

STEP 1: Talk to your doctor about FA Identified

- Before initiating genetic testing, your doctor can help determine if FA Identified is right for you

Even if your symptoms are mild or atypical for FA, genetic testing may still be appropriate.

STEP 2: Provide your sample

- If eligible, your provider will give you a genetic testing kit or you can provide your sample at your doctor's office
- You can provide a saliva, cheek swab (buccal), or blood sample—all are equally effective, so it's based on preference
- Follow all instructions in the testing kit carefully:
  - —If using a saliva or cheek swab, do not eat, drink, or chew gum for at least 30 minutes before collecting your sample
- If testing at home, be sure to mail the entire kit back using the provided FedEx envelope as soon as possible

STEP 3: Discuss your results with your doctor

- Results will be sent to your doctor within 3 weeks
- In rare cases, an additional test (called a reflex test) may be required, which could take an additional 2 to 3 weeks
- Your doctor can check the status of your results through the PreventionGenetics portal or by contacting PreventionGenetics directly

What happens if the test:

Confirms the FA diagnosis

Your doctor may recommend exploring treatment options to help address FA symptoms and genetic testing for family members to determine if they're at risk. **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.**

Rules out the FA diagnosis

Your doctor can use this information to exclude the FA diagnosis and prompt evaluation of other possible causes of your condition.

Identifies me as a carrier

Carriers have only one abnormal copy of the *FXN* gene and do not have FA, but there may be implications for family planning. Ask your doctor if genetic counseling is recommended to discuss potential next steps.

Take the Next Step: Talk to Your Doctor

Key questions to ask your doctor

- Do you recommend genetic testing for my symptoms?
- Am I eligible for the FA Identified no-charge genetic testing program?
- If genetic testing confirms that I have FA, what are the next steps?
- Should my family members consider genetic testing as well?
- Does your office have access to genetic counseling services? If so, would you recommend that I receive pre- and/or post-test counseling?

Key points to remember

Some ataxias are genetic—FA is the most common inherited form

FA is a rare and progressive genetic condition caused by a mutation in the *FXN* gene

Genetic testing is the only way to confirm an FA diagnosis

Eligible patients may qualify for no-cost genetic testing through FA Identified

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