

## What is Friedreich Ataxia (FRDA)?

Friedreich Ataxia (FRDA) is an inherited disease of the central nervous system. It was named after Nikolaus Friedreich, who first described it in 1863, and it was the first form of hereditary Ataxia to be distinguished from other forms of Ataxia.

## What are the symptoms of FRDA?

Difficulty with balance (disequilibrium), impaired coordination of the legs or arms, and thick or slurred speech. A speech disorder (dysarthria) is usually the first symptoms of Friedreich Ataxia.

Over time, problems with coordination and speech are likely to worsen. Curvature of the spine (kyphoscoliosis) and high arches in the feet (pes cavus) commonly develop. Affected individuals might notice difficulty knowing where their feet or hands are in space (impaired position sense) and they may develop weakness in the legs and hands.

Enlargement of the heart, irregular heartbeat, or other symptoms of heart trouble (cardiomyopathy) occur in many individuals with Friedreich Ataxia. Heart problems range from mild to severe. Diabetes mellitus is not uncommon.

Late in the course of the disease, about 10 percent of individuals with FRDA have a hearing loss, and a similar percentage develop loss of visual acuity or change in color vision. Another late-stage symptom in about 50 percent of affected people is difficulty with bladder control (incontinence).

## What causes FRDA?

Friedreich Ataxia is an inherited genetic disorder. It is caused by an abnormality of a single gene called the Frataxin (FXN) gene. The abnormality can be passed from generation to generation by family members who carry it.

Inherited diseases like FRDA occurs when one pair of the body's 20,000 genes does not work properly. (Genes are microscopic structures within the cells of our bodies that contain instructions for every feature we inherit from our parents. Two copies of each gene are inherited; one copy from the mother and one from the father.)

FRDA is autosomal recessive, which means that an individual only develops symptoms of the disease if both copies of his/her frataxin gene are not working properly. An individual who has one copy of an altered or nonfunctioning FXN gene does not develop any neurological symptoms and is called a carrier. For people who are carriers, the normal frataxin gene compensates for the nonfunctioning copy of the gene, however, a child whose parents are both carriers can inherit a "double dose" of the altered FXN gene and will therefore develop FRDA.

Most of the time carriers have no idea that they have an abnormal FXN gene because they do not have symptoms or medical problems that go along with being a carrier. It is often only when a child is diagnosed with FRDA that the parents learn, that they are both carriers. When both parents are carriers, each of their children has a 25 percent chance of having FRDA and a 50 percent chance of being a carrier.

## When do FRDA symptoms appear?

Males and females are equally likely to inherit the genes that cause FRDA. Symptoms usually begin between the age of 5 and 25 but occasionally appear in younger children or adults in their 30s or 40s.

## How Common is FRDA?

FRDA is the most common form of childhood onset Ataxia. In the United States, it is estimated that about 1 in 100 people is a carrier of the altered FXN gene and one out of every 20,000 to 50,000 is affected with Friedreich Ataxia. In some regions or ethnic groups this number might be a little higher or lower.

## **How is the Diagnosis made?**

When symptoms resembling those of FRDA appear it is important to receive a thorough medical evaluation by a neurologist. Generally, an evaluation will involve a physical exam and test to search for abnormalities in the brain and spinal cord. Many of these tests are done to rule out other possible causes of symptoms.

(Other possible causes might include nutritional deficiencies, infections, multiple sclerosis, herniated disc in the neck, stroke, brain and spinal cord tumors, and other degenerative diseases.)

Since the discovery of the FXN gene in 1996, it has been possible to make a specific diagnosis of FRDA by a gene test. In almost all cases, scientists can identify the abnormality in the frataxin protein, which is one of the thousands of proteins needed for the body to function properly. Levels of frataxin in the spinal cord and brain are much lower than normal in individuals with FRDA. However, it is more practical to test the FXN gene in blood cells than to measure frataxin protein levels in the nervous system.

Diagnosis of FRDA is made by genetic testing. In individuals with the typical clinical course, ancillary testing (EMG, MRI, CT) is unnecessary, though it may be useful in atypical patients. Appropriate specialists may be consulted such as a heart specialist, ophthalmologist, audiologist (hearing specialist), orthopedist (bone specialist), urologist, or endocrinologist (diabetes).

## **What happens after the diagnosis?**

It is helpful for patients and families with FRDA to undergo genetic counseling since they typically have questions about the chances that other family member will acquire the disease or be carriers of the abnormal FXN gene.

Questions about genetic testing can also be answered by a genetic counselor. An individual with FRDA should find a physician who will follow him or her on a regular basis to help address the neurologic changes that are likely to occur over the course of the disease, to anticipate and screen for possible complications (such as diabetes and heart disease), and to make appropriate referrals to other specialists as needed, including physical, occupational or speech therapists.

## **What kind of support is available for people with FRDA and their Families?**

The National Ataxia Foundation (NAF) is committed to providing information and education about Ataxia, support groups for those affected by Ataxia, and promoting and funding research to find the cause for the various forms of Ataxia, better treatments, and, hopefully someday, a cure. NAF has been at the forefront funding promising worldwide research to find answers.

As Ataxia research moves into the clinical phase, pharmaceutical companies will begin recruiting participants for clinical trials. Individuals with Ataxia or who are at-risk for Ataxia are encouraged to enroll in the CoRDS Ataxia Patient Registry. To access the Registry, go to NAF's website [www.ataxia.org](http://www.ataxia.org) and click on the "Enroll in the Patient Registry" tab and follow the directions on the CoRDS website. Individuals with Friedreich's Ataxia can also register in the [FA Global Registry](#).

NAF provides accurate information for you, your family, and your physician about Ataxia. Please visit the NAF website at [www.ataxia.org](http://www.ataxia.org) for additional information, including a listing of ataxia support groups, physicians who treat Ataxia, social networks, and more. For questions contact the NAF directly at 763/553-0020 or [naf@ataxia.org](mailto:naf@ataxia.org).