

Friedreich's Ataxia (FA) Facts

Friedreich's ataxia, what it is?

Friedreich's ataxia is a severe disease which was described for the first time by the German neurologist Nicolas Friedreich in 1881.

It is a progressive disorder, mainly affecting the nervous system. FA affecting about 1-2 people in 40,000 in Canada. Males and females are affected equally.

What happens to people with FA?

FA symptoms usually start early, with the average age of onset being around 15 years old. However, symptoms can start to show in very young children or in older adults. The early symptoms can be quite varied but usually involve poor co-ordination and clumsiness, followed by balance problems when walking. As FA progresses, speech generally becomes slurred. The rate the disease progresses varies considerably between patients, however. After an average of 10 years from the onset of symptoms, people with FA usually find that their legs become weak and difficult to co-ordinate and they may need to use a wheelchair. They may also find it difficult to write.

FA affects different people in different ways. Some people may also experience one or more of the following symptoms: hypertrophic cardiomyopathy (thickening of the heart wall that causes weakening of the heart), scoliosis (curvature of the spine), pes cavus (excessively arched foot), diabetes or impaired glucose tolerance, nystagmus (rapid involuntary movements of eyes), swallowing difficulties, reduced vision, reduced hearing, cold feet due to decline in muscle activity.

While FA results in physical disability it is not generally thought to affect people's intellect or mental capabilities. But there are often emotional aspects of coming to terms with FA and the associated disability, and these also vary from person to person. People with FA may become depressed. In general, life expectancy is shortened but it is not possible to say by how much. Like other disabilities FA does have an impact on people's lives; however many people with FA lead full, active and purposeful lives. For example there are many people with FA who go to university, work, travel the world or start families of their own.

FA affects people in different ways; symptoms can vary in terms of severity and types.

How do you get FA?

FA is inherited. If you have FA you have inherited one faulty copy of the FA gene from your mother and one from your father. This is called recessive inheritance. You can get FA even if your parents do not have ataxia themselves, as they will only be carriers of FA. Carriers only have one copy of the faulty gene instead of two. If both parents are carriers the risk of having a child with FA is one in four and the risk of having a carrier child is one in two. If you have FA the only way in which you can have a child with FA is if the other parent has FA or is a carrier of FA. The number of people that have a single faulty FA gene (carrier frequency) in the Canada population is about one in 80-100. If one parent has FA and the other is a carrier there is a one in two risk of having a child with FA.



CAFA
Canadian
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for Familial
Ataxias

Founded by Claude St-Jean

(Formerly
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Diagnosis

A genetic test is available that will provide a firm diagnosis of FA. It involves taking a blood sample and can be arranged through your doctor. Prenatal diagnosis is available if parents already have a child with FA and are considering having further children. This can tell you if your child will develop FA, will be a carrier, or will be completely free of the faulty gene.

What causes FA?

FA is caused by a defect in both copies of the FA genes. In the majority of patients (96%) this defect is an expansion in the size of the gene in both copies of the gene. Both normal and defective FA genes produce a protein called frataxin. But the defective gene fails to produce enough of the protein. This reduction in frataxin in turn leads to the disorder. Frataxin is essential for life. It is found in specific compartments within cells called mitochondria, which produce the energy that cells need to survive and work properly. In people with FA there is an accumulation of iron and potentially toxic chemicals known as “free radicals” in the mitochondria. These will damage the mitochondria and other parts of the cells. The damage reduces the energy supply to the cells, which then causes them not to work properly and eventually to die. Frataxin is thought to play a role in regulating the iron levels in the mitochondria or it could act to protect the cell from damage by free radicals (a “free radical scavenger” or antioxidant). Research is still underway to understand in detail what happens in the bodies of people with FA in order to develop treatments.

What can be done?

Health Canada approved Catena® for treatment of Friedreich’s ataxia to address the needs of Canada’s several hundred patients with this rare and severely progressive neurodegenerative disease. Until very recently there was no medication available to improve the condition of FA patients. Idebenone as well as Catena, which is its new high dose formulation, seem to cause an improvement at the neurological and cardiac levels. Two ongoing studies have yet to prove their effectiveness for all patients. Hopefully other drugs will appear on the market and contribute to ease the clinical symptoms of a maximal number of patients.

Multiple ongoing studies on inhibitors of the histone deacetylase enzyme (HDACIs) are trying to demonstrate their effectiveness in several types of cancer or neurodegenerative diseases. It seems that only a specific group of HDACIs is able to significantly increase the level of frataxin in human body cells, particularly in immature white blood cells as well as in organs singled out by Friedreich’s ataxia.

How can FA be managed?

A number of symptoms of FA can be treated. The early assessment and regular follow-up of heart problems by a cardiologist is important to prevent complications. Diabetes may sometimes be treated with insulin therapy. Orthopaedic surgery can help correct problems of the spine and feet. If muscle spasms are a problem, medications such as baclofen can possibly help. If the emotional effects of FA include depression this can also be treated. Any treatment must be discussed with a family doctor or a neurologist first. Physiotherapy and exercise such as swimming are important to work against the loss of strength and preserve mobility. A speech



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therapist can help you with problems involving speech, swallowing, coughing and choking. A full assessment is required, and this can be arranged by your doctor.

If speech becomes too difficult then the speech therapist can advise you on the best communication aids. An occupational therapist can also be helpful, for example with home adaptations.

What next?

Although there is no hiding the fact that FA does impact on people's lives, it does not have to prevent ataxic people from having a full, active and enjoyable life.

Canadian Association for Familial Ataxias (CAFA) – Claude St-Jean Foundation is here to support you, so do contact us if there is anything that we can help you with. Many of our members with FA or their parents would also be happy to share their experiences.

For more specific advice, information or support, please contact us at CAFA Office.

A list of medical and scientific references is available from the Office on request.

How you can help to fund the research?

Organize a fundraising event to benefit CAFA: evening event, show, supper fundraiser, draw, marathon, golf tournament, etc. Let us be a part of your project.

Promote yourself through our cause. Artists, designers, inventors! Organize a show, event or art exhibit and enjoy increased visibility by supporting CAFA's cause.

Make a donation of goods or service.

Are you thinking of getting rid of your old car? Give it to CAFA and receive a tax purpose receipt.

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Some extracts are taken from Ataxia UK web site