



ATAVIA

Ataxia UK

Friedreich's ataxia the facts

ATAVIA



Professor Marios Hadjivassiliou of the Sheffield Specialist ataxia centre.

Introduction

The information in this leaflet has been reviewed by ataxia expert neurologist **Professor Marios Hadjivassiliou** (Sheffield Teaching Hospitals NHS Foundation Trust). We have also received input from a number of people who have ataxia.

This leaflet contains information about **Friedreich's ataxia** and is aimed at everyone affected by it. You may have been diagnosed with Friedreich's ataxia yourself, you may be a parent, relative, friend or you may be a carer for someone with Friedreich's ataxia and wish to know more.

If you have any questions or concerns after reading this leaflet, please contact the Ataxia UK helpline:

0800 995 6037

Disclaimer

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CONTENTS

What is Friedreich's ataxia? ————— 4

What are the symptoms of Friedreich's ataxia? ——— 6

When and how do symptoms start? ————— 8

How do you get Friedreich's ataxia? ————— 9

What causes Friedreich's ataxia? ————— 10

How is Friedreich's ataxia diagnosed? ————— 12

How common is Friedreich's ataxia? ————— 13

Management of Friedreich's ataxia — 14

Monitoring progress ————— 15

Heart problems ————— 15

Curved spine (scoliosis) ————— 16

High arches and turned-in feet ————— 16

Eye symptoms ————— 17

Swallowing difficulties ————— 17

Muscle spasms ————— 17

Diabetes ————— 18

Reduced hearing ————— 18

Referral to therapists ————— 19

What will the future be like? ————— 20

References ————— 23

What is Friedreich's ataxia?





**Dr Nikolaus
Friedreich**

The word **ataxia** means *lack of co-ordination*. There are many different causes and types of ataxia. **Friedreich's ataxia** is one type and it takes its name from **Dr Nikolaus Friedreich** who was the first person to describe this condition.

Friedreich's ataxia is a progressive condition, which means it gets worse over time. You may also hear the term **degenerative** meaning the same thing. Friedreich's ataxia is often called **FA** (or **FRDA**) for short.

Some of the main symptoms experienced by people with FA happen because certain nerve cells in the body are damaged and eventually die. Sensory nerve cells in the spinal cord and nerve cells that connect the spinal cord to other parts of the brain are mostly affected. Other problems arise because cells are also affected in different parts of the body such as the heart (causing cardiomyopathy) and pancreas (causing diabetes).

Sam Merchant, who has FA, is an active fundraiser for Ataxia UK



What are the symptoms of Friedreich's ataxia?

It is important to remember that FA affects individuals in a variety of ways and different degrees.

Symptoms may be worse in some people than in others and some symptoms may not appear at all.

Millie Mae, who was diagnosed with FA at the age of 8.





Ant Brown, who was diagnosed with FA when he was 12, is now heavily involved in the **South West Support Group**.

Main symptoms

- **Poor balance and coordination** (ataxia)
- **Difficulty speaking** (dysarthria)
- **Swallowing difficulties** (dysphagia)
- **Curved spine** (scoliosis)
- **Heart problems** (cardiomyopathy)
- **Reduced sensation in arms and legs**
- **Difficulty in handwriting**

Other common symptoms

- **Those associated with diabetes**
- **Muscle spasms** (usually affecting legs)
- **High arches of the feet** (pes cavus)
- **Feet turned inwards at the ankles** (talipes equinovarus or 'clubfoot')
- **Involuntary eye movements** (nystagmus)
- **Cold, discoloured feet or lower limbs**
- **Poor bladder control**
- **Breathing difficulties** (eg. sleep apnoea)
- **Hearing problems**
- **Reduced vision**
- **Fatigue**

As with other chronic conditions, people with FA can sometimes be affected by mood disorders such as depression. These are treatable so it is important to discuss them with a doctor.

When and how do symptoms start?

Symptoms of FA usually start before the age of 25. On average, people are around 12-15 years old when symptoms first appear. However, symptoms can sometimes begin in very young children or in older adults (known as late onset FA). It is very rare for symptoms to start after the age of 40.

The early symptoms usually involve poor co-ordination and clumsiness, followed by balance problems when walking. Speech generally becomes slurred as Friedreich's ataxia progresses but how fast this happens varies a lot between different people. After an average of ten years from when symptoms begin, people with FA will probably need to use a wheelchair. It may also be difficult to write.

Irish Paralympic medalist, **Helen Kearney** was diagnosed with FA in 2001.





How do you get Friedreich's ataxia?

Friedreich's ataxia is an inherited condition, which means it is passed on from parent to child. Everyone has two copies of the gene involved in FA. If you have FA, you have inherited two faulty copies, one from your mother and one from your father. This is called **recessive inheritance**.

You can get FA even if your parents don't have the condition. People who have just one faulty gene are known as **carriers**. They will never develop symptoms themselves, but they can pass on the faulty gene to their child.

If both parents are carriers:

- the chance of having a carrier child is **1 in 2**;
- the chance of having a child with FA is **1 in 4**; and
- the chance of having a child who is unaffected is **1 in 4**.

For more information on this see Ataxia UK's publication: **Ataxia: What's that?**

After the genetic diagnosis is made, it is common practice for the person with Friedreich's ataxia and their family to be referred to the local clinical genetics service, so that the above information regarding inheritance and any risk to the next generation can be addressed in detail.

What causes Friedreich's ataxia?

27 year old **Iain Fryatt** has FA.

But he didn't let it stop him climbing to the top of Mount Kilimanjaro in his wheelchair.

Friedreich's ataxia is caused by a fault in a gene called **frataxin**. All genes are made up of molecules called **nucleotides** that are held together in a chain. Each nucleotide is one of four types, known as A, T, C or G. People have two copies of each gene.

In most people with FA, both copies of the frataxin gene are longer than usual because the sequence G-A-A is repeated too many times. A small minority (2-4%) of people with FA have the G-A-A fault in one copy of the frataxin gene and another type of fault in their other copy. Both the normal and faulty frataxin genes produce a protein which is also called frataxin; however, the faulty frataxin gene gets 'switched off' and therefore doesn't produce as much frataxin protein. This shortage of frataxin protein is what leads to the symptoms of FA.



Every cell in the body contains the frataxin protein, inside specific cell compartments called **mitochondria**. Mitochondria are the 'power plants' that produce the energy cells need to survive. Frataxin has a role in the process of energy production. When there is less frataxin, this process is disrupted and it leads to a number of consequences. For example, toxic chemicals build up within the mitochondria and cause damage – known as **oxidative stress**. There is a reduction in the energy supply to cells. Without enough energy, the cells can't work properly and eventually they die. An accumulation of iron also occurs within the mitochondria, which again is problematic for cells.

Research is taking place to understand the detail of what happens in people with FA in order to develop treatments. A number of treatment options are being explored that address these problems caused by the faulty frataxin (*see page 21 for current research*).



Trainer and workshop facilitator, **Toyah Wordsworth**, was diagnosed with FA as a child.

How is Friedreich's ataxia diagnosed?

A genetic test can provide a firm diagnosis of FA. It involves taking a blood sample and it is usually requested by your neurologist or geneticist.

If you are thinking of having a baby and you have Friedreich's ataxia, it is possible for your partner to be tested to see if they are a carrier of the faulty gene. If they are not a carrier, then it is extremely unlikely that the child will develop FA. It is also possible to have a test done before the pregnancy (if you are having **in vitro fertilisation** or **IVF**) or a prenatal test during pregnancy. The tests can tell you whether your child will develop FA or will be a carrier.

You will need to talk to a **genetic counsellor** or **neurologist** with expertise in genetic counselling before having any genetic test, so that you can talk through the possible results and how they might affect you. Your neurologist or GP can refer you.

Nigel Kilvington was diagnosed with FA in his late 30s.



How common is Friedreich's ataxia?

FA is the most common type of **inherited ataxia**, although it is still a rare condition. Estimates from a European study of Caucasian people suggest that around one person in 30,000 has FA. This would mean there are about 2000 people with FA in the UK. However, FA is rarer in people of sub-Saharan African origin and in people who originate from the Far East. In a European study, numbers were found to be high in parts of Spain, France and Ireland. Men and women are equally affected.



Chris Hobson, who is now 30, developed FA when he was 12.

How to manage Friedreich's ataxia?



There is currently no cure for Friedreich's ataxia, but there are a number of treatments and recommendations available to help with the symptoms. These are outlined below.



Monitoring progress

People with Friedreich's ataxia should see a neurologist at least once a year. The neurologist can monitor the condition and provide help with any new problems, if there are any. It is also an opportunity to find out about any new medical advances. Ataxia UK has accredited **Specialist Ataxia Centres** with expertise in monitoring progress and managing symptoms of people with ataxia. A referral can be arranged via the GP (*see Ataxia UK's website for details*).



Heart problems

About two thirds of people with FA have heart problems (**cardiomyopathy**). It is therefore essential for people with Friedreich's ataxia to see a **cardiologist (heart doctor)** in order to diagnose or anticipate any problems as early as possible. Screening should happen once every two years if there are no symptoms of heart trouble or once a year if heart symptoms have already been diagnosed. Some people may need medication to control an irregular heartbeat or to prevent heart failure, depending on their symptoms. The cardiologist usually works very closely with the neurologist in monitoring the person with FA.



Curved spine (scoliosis)

The neurologist (adult or paediatric) should check for any signs of **curved spine** at the yearly appointment. This is especially important for children as **scoliosis** can in the future cause severe pain, discomfort and respiratory problems. Depending on the severity, treatment may be required and such treatment is best done at an early age. If necessary a referral to a spinal unit will be made to see an **orthopaedic surgeon** and have an x-ray image taken. Physiotherapy or wearing a brace around the body to keep the spine straight may be recommended. In severe cases the spinal surgeon may suggest having an operation to straighten the spine.



High arches and turned-in feet

Regular physiotherapy may prevent having permanent **'turned in' feet**. Wearing custom-made insoles for shoes can help reduce the pain caused by high arches. The GP or neurologist can make a referral to an **orthotist** for these to be made. In severe cases, corrective surgery can be done by **orthopaedic surgeons** who specialise in foot surgery.



Eye symptoms

Involuntary eye movements can sometimes cause an unpleasant feeling that the world you see is moving (**oscillopsia**). If so, there are several medications that can sometimes help.

The ability to see detail clearly may become worse as Friedreich's ataxia progresses. This is because of degeneration of the optic nerves. There is currently no treatment, but it is possible to become registered as partially sighted and the individual may be entitled to benefits or practical support, via social services.



Swallowing difficulties

If people are experiencing coughing or choking while eating and drinking, it is important to get a referral to a **speech and language therapist** who can give advice on ways to overcome such problems. For example, they might suggest changing the texture of food and drink, tucking in the chin when swallowing or adjusting the way someone sits.



Muscle spasms

A number of people with FA suffer from **muscle spasms** that can be painful and cause discomfort. There are medications that can help alleviate symptoms, so it is important to discuss this with the neurologist.



Diabetes

Most people with Friedreich's ataxia who develop **diabetes** will need to be treated with standard diabetes treatment involving the hormone **insulin**. This will lower the amount of blood sugar and help to prevent complications.



Reduced hearing

Some people with FA have problems with hearing, especially in situations with background noise. This is a neurological problem and is due to the quality of the sound that can be detected rather than the volume. For this reason, traditional hearing aids that amplify the sound do not often help, although it is sometimes worth trying them. A referral to an **audiologist** is recommended for advice on treatments. New hearing devices are being trialled that improve the quality of the sounds and will hopefully be more useful in FA.



Referral to therapists

A **physiotherapist** can be very beneficial and suggest balance exercise to improve mobility and maintain muscle strength. Physiotherapy and exercise such as swimming may also prevent loss of strength and preserve mobility. A **speech and language therapist** can help with problems involving slurred speech and, if needed, with time can advise on communication aids. An **occupational therapist** can also be helpful, for example, with home adaptations, teaching strategies for daily activities or wheelchair assessments.



Jonathan Kay, 34,
and his friend, Daniel,
celebrate after their
charity skydive
- courtesy of Ataxia
UK's **Jerry Farr Travel**
Fellowship

What will
the future
be like?



Current research

Although there is as yet no approved treatment for FA, there are many different researchers around the world exploring ways of treating the condition. Studies are exploring **anti-oxidants**, drugs that enhance function of the **mitochondria**, drugs increasing levels of **frataxin protein**, **gene therapy** and **stem cell therapy** approaches. Many of these approaches have shown promise in studies involving animal models of FA and have progressed to the stage of testing treatments in trials involving people with FA. Some pilot studies testing the effect over a few months have moved to longer trials lasting one year or over.

Ataxia UK is actively involved in funding and supporting the research effort in many ways and we are committed to finding treatments and ultimately cures. If you are interested in keeping updated on research developments or in hearing about opportunities to take part in research studies, please join Ataxia UK. *For more information on research visit the Ataxia UK website research section.*

Research into FA is being undertaken across the globe.



What next?

Although there is no hiding the fact that FA does impact on people's lives, it does not have to prevent people from leading a full, active and enjoyable life. Ataxia UK is here to give support to anyone affected by ataxia, so do contact us if there is anything we can help with. Joining Ataxia UK is free and we offer a number of support services as well as being committed to research and finding treatments and, ultimately, a cure. Please do join Ataxia UK and support us in our work.

For more specific advice, information or support, or if you would like a large print version of this leaflet, please contact our helpline (**0800 995 6037**). We also have a number of other publications that may be useful including information on living with ataxia in the leaflet: **ataxia: what's that?** and **Ataxia Guidelines for healthcare professionals**. It may be helpful to provide copies of these guidelines to the GP, neurologist or therapist. These can be found on the Ataxia UK website: www.ataxia.org.uk.

Ataxia UK Trustee,
Richard Brown, was
diagnosed with FA at
the age of 15.



References

The following papers were consulted in writing this leaflet:

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