Introduction

This leaflet has been written to provide people affected by Friedreich’s ataxia (FA) with factual information about the condition. You may have been diagnosed with FA yourself, you may be caring for someone with FA, or you may be a parent, relative or friend and wish to know more. We hope that this leaflet will help you by explaining some of the facts. It is important to note that FA affects people in different ways; symptoms can vary in terms of severity and types. If after reading this leaflet you have any questions or concerns, please contact us.

What is Friedreich’s ataxia?

Dr Nicolaus Friedreich was the first person to describe the symptoms of this kind of ataxia and so it has taken his name. There are many different types of ataxia, some of which are inherited. The word ataxia simply means ‘lack of co-ordination’. Friedreich’s ataxia is a progressive disorder mainly affecting the nervous system. It is often called FA (or sometimes FRDA) for short.

The main problems experienced by people with FA are caused because certain nerve cells in the body become damaged and eventually die. The nerves that are mainly affected are those of the spinal cord and those that connect the spinal cord to other parts of the body, such as the arms and legs. Other cells in the body are also affected including the heart and pancreas, accounting for other symptoms associated with FA.

How many people have FA?

Although it is the most common of the inherited ataxias, it is still rare, affecting about 1-2 people in 50,000 in the UK. Males and females are affected equally.

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**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 vary 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. Although some people do die young, there are people with FA in middle and old age.
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.

**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 (see our Genetics leaflet or website for more information). 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 chance of having a child with FA is one in four and the chance 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 UK population is about one in 80-100. If one parent has FA and the other is a carrier there is a one in two chance of having a child with FA.

**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. In about 4% of cases patients have one expanded gene and a different defect in the other 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?**

There is currently no proven treatment available for FA. However, there have been a number of treatment trials using antioxidants (currently idebenone, Vitamin E and Coenzyme Q10). It is hoped that these antioxidants will reduce the damage by free radicals, increase the energy supply to cells and so slow or stop progression of the ataxia. Preliminary results with these therapies have been promising.

**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, and nystagmus might be treated with botulinum toxin injections. 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 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 you from having a full, active and enjoyable life. We are 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, or if you would like a large print version of this leaflet, please contact us at the Ataxia UK Office.

A list of medical and scientific references is available from the Office on request.
This leaflet was produced by Ataxia UK. The following people also contributed to the production of this leaflet: Dr de Silva (Consultant Neurologist, Oldchurch Hospital), Dr Cooper (Senior Lecturer in Clinical Neurosciences, Royal Free and University College Medical School London), Mr Law, and Mr and Mrs Ashton (members of Ataxia UK).

Ataxia UK would like to thank NM Rothchild & Sons Limited for their generous donation towards the cost of producing this leaflet.

Ataxia UK, Lincoln House, Kennington Park, 1-3 Brixton Road, London SW9 6DE
www.ataxia.org.uk  helpline@ataxia.org.uk
Tel: 020 7582 1444  Helpline: 0845 644 0606

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