

Ataxia  
UK



Information on  
Cerebellar ataxia

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Rose at her  
computer

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## Introduction

This leaflet is for you if:

- you have been diagnosed with cerebellar ataxia
- you are caring for someone with cerebellar ataxia
- you are a parent, relative or friend of someone with cerebellar ataxia and wish to know more.

We hope that this leaflet will answer most of your questions but if after reading it you have any questions or concerns, please contact us.

## What is cerebellar ataxia?

There are many different types of cerebellar ataxia, not just one, and in this leaflet we refer to them all as CA. Some have been found only in a few families in specific countries; others have been identified throughout the world. It is fair to say that any kind of ataxia is a rare disorder. But you are not alone, even though you may feel that way at times. Many people do not know about ataxia, and you may find it helpful to give them a copy of our leaflet "Ataxia – The Facts" to help them understand ataxia better and how it affects you.

The word “cerebellar” comes from “cerebellum”, part of the brain that, among other things, controls balance and co-ordination. The word ataxia simply means “lack of co-ordination”. So the cerebellar ataxias are disorders in which the nervous system (including the cerebellum) is affected, causing problems such as unsteadiness and lack of co-ordination. They are all very rare disorders, with probably only a few thousand people affected in the UK, though more people are affected world-wide.

Ataxia can also be a symptom of other conditions, such as multiple sclerosis or cerebral palsy. If you have one of these conditions we can help you live with your ataxia, but for other symptoms you may find it more helpful to contact the organisation for the relevant disorder (e.g. Multiple Sclerosis Society – [www.mssociety.org.uk](http://www.mssociety.org.uk) or Scope – [www.scope.org.uk](http://www.scope.org.uk)) for more detailed information.

## What are the symptoms?

Most of the cerebellar ataxias are progressive, so will get worse with time. The speed at which the ataxia progresses varies between different types of ataxias and between individuals, but is normally very slow, and changes take place over many years. The age at which symptoms begin also varies between the types of cerebellar ataxias, for example some tend to start in childhood but others tend to start later on in

life. There are some types of CA that are not progressive.

The symptoms of CA include poor co-ordination of the arms and legs. As the disorder progresses, walking may become difficult or even impossible, so that those affected may need a wheelchair. It is important to note that there is a very large variation in the severity of the symptoms. Some people are affected very mildly: for example, they only experience slight balance problems. Others experience a number of different symptoms more severely. The symptoms of CA may also include:

- Slurred speech (dysarthria)
- Problems with swallowing (which may appear as coughing or choking)
- Tremors
- Nystagmus (rapid involuntary movements of the eyes)
- Problems with sight
- Problems with hearing
- Cold feet

There may also be other symptoms that are associated with a specific type of CA. Like other disabilities CA does have an impact on people's lives; however many people with CA lead full, active and purposeful lives. Many people with the disorder go to university, work, bring up families, socialise or travel the world.

## What causes cerebellar ataxia?

Some types of cerebellar ataxia are inherited and some are not. The non-inherited types, also known as sporadic cerebellar ataxias, can have a number of causes. All types of cerebellar ataxia are described in more detail below.

### **Inherited cerebellar ataxias**

The inherited cerebellar ataxias can be divided into four groups, depending on how they are inherited. The disorder can either be inherited in an autosomal dominant way or an autosomal recessive way; it can be transmitted only from the maternal line (mitochondrial ataxias), or rarely can be X-linked (only males are affected). For further explanation on the types of inheritance see the Ataxia UK leaflet "Genetics and ataxia" or the Ataxia UK website ([www.ataxia.org.uk](http://www.ataxia.org.uk)). The inherited ataxias involve a defect in a gene, which can be passed on through the generations. In some types of inherited cerebellar ataxia the severity of the disorder can increase as it gets passed down a generation, and the age of onset can get younger. This is called "anticipation".

**Autosomal dominant inheritance** – Ataxia is caused by having one copy of the faulty gene that has been inherited from only one of the parents. If you have ataxia of this kind there is a one in two chance of passing the ataxia to your child. Examples of ataxias inherited in this way:

- Spinocerebellar ataxias (SCAs)

A number of spinocerebellar ataxias have been identified. These all have an abnormality in a gene. As each gene abnormality has been found it has been given a number, for example SCA1, SCA2, SCA3 and so on. Although the cause is different they have many symptoms in common and sometimes it is only possible to tell the difference between them by genetic testing. At the moment we know of 20 different SCAs. Genetic tests are not yet available for all of them. Currently tests are only available for SCAs 1, 2, 3, 6, 7, 10, 12 and 17.
- Dentatorubral-pallidoluysian atrophy (DRPLA)

DRPLA has some similarities with Huntington's disease. Symptoms of DRPLA include sudden muscle spasms and epilepsy.
- Episodic ataxia type 1 (EA-1)

EA-1 differs from the majority of ataxias because it is characterised by short attacks of inco-ordination and dysarthria (slurred speech) that generally last minutes. Also EA-1 is generally a non-progressive disorder, but some elderly people show slight permanent ataxia and tremor. Attacks can occur spontaneously or they can be triggered by rapid sudden movements, or if one is startled. Anxiety and fatigue also increase the susceptibility to an attack.

- Episodic ataxia type 2 (EA-2)

EA-2 involves episodes of ataxia that last hours to days. It is a rare disorder, representing less than 1% of the inherited ataxias. The medication acetazolamide may prevent or lessen episodes of ataxia. Please note that the use of any medicines must always be discussed with a doctor. As stress often triggers attacks, stress management techniques (e.g. meditation) can be helpful in controlling symptoms. For more information on controlling stress contact your doctor.

**Autosomal recessive inheritance** – In these cases, the ataxia is caused by having two copies of a faulty gene, one inherited from each parent. A child can be born with ataxia of this kind if both parents have one faulty copy of the gene. This makes the parents carriers of the disorder, although they do not have ataxia themselves. If the two parents are carriers there is a one in four chance of having a child with ataxia and a one in two chance of having a child who does not have ataxia but who carries the faulty gene and may pass it on in their turn. In other words, that child is a carrier. Examples of some of the ataxias inherited in this way are:

- Friedreich's ataxia – the most common of the inherited ataxias (see the Ataxia UK Friedreich's ataxia leaflet for more details)
- Ataxia telangiectasia (contact A-T Society for more details – [www.atsociety.org.uk](http://www.atsociety.org.uk))

- Ataxia with Isolated Vitamin E deficiency
- Abetalipoproteinemia
- Cerebellar ataxia with muscle Coenzyme Q10 deficiency
- Early-onset cerebellar ataxia with retained tendon reflexes
- Infantile onset spinocerebellar ataxia
- Marinesco-Sjogren syndrome
- Spastic ataxia of Charlevoix-Saguenay
- Ataxia with oculomotor apraxia

There are numerous other types of recessive ataxias, some only identified in a few families.



Frankie and  
his brother  
Ryan

**Mitochondrial disorders** – These involve mutations in the genes that are found in the mitochondria, the energy-producing compartments of cells. As each person inherits their mitochondria from their mother, this means that these disorders can only be passed down the maternal line. Most of the genes found in the mitochondria are involved in the production of energy, so generally mitochondrial disorders result from an incapacity to produce sufficient energy within cells, preventing them from doing normal functions. Some mitochondrial disorders have ataxia as a main symptom. Examples of mitochondrial ataxia disorders are:

- NARP (neuropathy, ataxia, and retinitis pigmentosa)
- MELAS (mitochondrial encephalomyopathy, lactic acidosis with stroke-like episodes)
- Myoclonus epilepsy with ragged red fibres (MERRF)

**X-linked inherited ataxias** – Very rarely, the gene for ataxia can be carried on the X chromosome. This usually means that only males are affected. Females can be carriers and transmit the disorder to their sons.

### **Non-inherited cerebellar ataxias**

Some people who have ataxia do not have a history of ataxia in the family. These people may either have a recessively inherited ataxia that has not been identified yet or they may have a non-inherited form of ataxia. If the cause of the ataxia is not known it is sometimes called

idiopathic ataxia. For example, a number of people are diagnosed as having “idiopathic late onset cerebellar ataxia”. This condition occurs later on in life and is idiopathic (i.e. no known cause). People can be diagnosed with this if there is lack of evidence of a genetic or other cause. It is often slowly progressive and has few additional symptoms. Examples of non-inherited ataxia include:

- Multiple system atrophy  
This is a condition that occurs later on in life. It is a progressive cerebellar ataxia, and the symptoms are sometimes similar to Parkinson’s disease. The Sarah Matheson Trust is a charity that provides support and information specifically for this condition ([www.msaweb.co.uk](http://www.msaweb.co.uk)).
- Gluten ataxia  
Researchers have found that some people with idiopathic ataxia have a sensitivity to gluten (found in grains such as wheat and barley). This may be the cause of their ataxia, and research is underway to study this further and to see whether a gluten-free diet is beneficial.
- Ataxia due to trauma or toxic exposure  
A number of people have ataxia that is related to injury to their brain, either from exposure to toxic substances (which can include alcohol in large enough quantities over a long period of time), head injury or brain surgery. Viral infections or tumours can also cause ataxia.

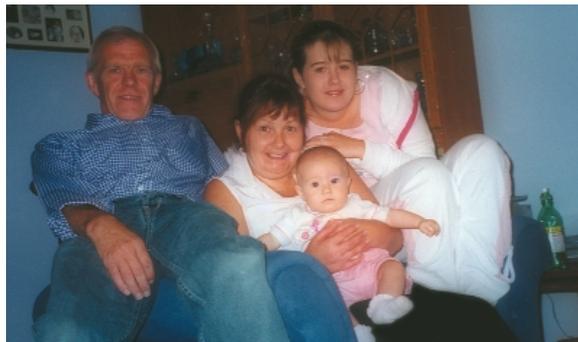
- Ataxia due to cerebellar malformations

Ataxia can also be caused by a malformation of the cerebellum that mostly occurs before a baby is born. The symptoms start in childhood. There are many different disorders, some are inherited and some are caused by external factors. Examples of cerebellar malformations are the Dandy-Walker malformation and Chiari malformation.

## How fast does the ataxia progress?

## What will happen when?

These are the hardest questions of all to answer. As a very rough guide, the earlier the ataxia starts, the faster it progresses, but it is impossible to predict what will happen in a particular case. We need research to find the answers to these questions.



Liz and her family

## Non-progressive ataxias

The majority of cerebellar ataxias that Ataxia UK deals with are progressive. There are some types that are non-progressive, and we still aim to help people with the symptoms of these types of ataxia.

For example disorders that involve malformations of the cerebellum that occur before a baby is born can be non-progressive. When children get CA as a result of a virus such as chickenpox a full recovery generally occurs within a few months. Ataxia due to brain damage or tumours may also be non-progressive.

## Are mental abilities affected?

Often the effects of CA are confined to impaired co-ordination. Some forms of CA can affect other parts of the brain, including those parts involved in intellectual function. There are also emotional aspects of coming to terms with CA, and these can vary from person to person.

In a few types of CA, research is now beginning to show that the cerebellum or other parts of the brain affected by CA may be involved in emotions, thought and behaviour. This may result in people with CA experiencing mood disorders, such as depression. This is understandable, and treatable, so they should see their doctor.

## Diagnosis

It is sometimes difficult to give a firm diagnosis of the specific type of CA you have, as there are many different disorders that appear very similar. A neurologist may need to do extensive tests to find out what a patient has. These include:

- Patient history  
Helps identify if the ataxia is caused by a tumour or alcohol poisoning, for example.
- Brain scans  
MRI brain scan (magnetic resonance imaging) will give an image of the cerebellum and other parts of the brain and show if it is damaged. For example it can sometimes be used to distinguish between Multiple Sclerosis and CA.
- Family history  
Helps to determine whether the patient has an inherited CA. If parents and grandparents also have ataxia then it is likely that the patient has an autosomally dominant inherited CA. If the parents are not affected but more than one of the children has CA it suggests that it is inherited recessively. Even if no-one else in the family is affected, this does not necessarily mean that CA is not inherited; for example an affected parent may have died before the onset of ataxia symptoms.
- Genetic tests  
Available for some of the inherited cerebellar ataxias, e.g. Friedreich's ataxia, SCAs 1, 2, 3, 6, 7, 10, 12, 17 and

DRPLA. If the results are positive, then it can generally be considered a definite diagnosis. If the results are negative for all these tests, this may mean that you have a type of inherited CA for which the gene has not been found or a test is not yet available.

- Other laboratory tests  
e.g. testing for Vitamin E levels in the blood to see whether a patient has Ataxia with Isolated Vitamin E deficiency or abetalipoproteinemia. People with these conditions have low levels of Vitamin E, and this can be treated by Vitamin E supplements.

Some types of CA have specific characteristics that make them distinguishable from others, e.g. SCA7 (retinal degeneration causing problems with eyesight).

## What are the treatments?

There is currently no cure available for the majority of the cerebellar ataxias. If you have been diagnosed with Ataxia with Isolated Vitamin E deficiency or abetalipoproteinemia these can be treated by Vitamin E supplements and a specialised diet. The newly recognised disorder called cerebellar ataxia with Coenzyme Q10 deficiency may be treated with Coenzyme Q10 supplements. In addition, a number of treatment trials have been ongoing in people with Friedreich's ataxia (see Friedreich's ataxia leaflet).

In the absence of a cure there are a number of treatments available to help with the symptoms of CA. If muscle spasms are a problem, medications such as baclofen may help. Medications are also available for tremors or bladder problems. People with abnormal eye movements sometimes benefit from botulinum toxin injections, or with the use of prisms incorporated into glasses. If the emotional effects of cerebellar ataxia include depression this can also be treated. Any medications should always be discussed with a doctor or neurologist first.

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John in his role as a local councillor

Physiotherapy and exercise such as swimming may prevent loss of strength and preserve mobility. A speech therapist can help with problems involving speech, swallowing, coughing and choking. If speech becomes too difficult then the speech therapist can also advise on the best communication aids. An occupational therapist can also be helpful, for example with home adaptations, wheelchair assessments and general well-being.

## Research

Research into treatments for many types of CA is taking place throughout the world, including the UK. One important way in which you can help research is to ask your doctor to tell you exactly what kind of cerebellar ataxia you have, and to let us know. This will help us to put together groups of people with the same kind of ataxia for potential future research projects. We realise that this is very personal information, which Ataxia UK will hold wholly confidentially and in accordance with the Data Protection Act 1998. Your details will not be disclosed to anyone without your permission.

## What next?

Although there is no hiding the fact that CA 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 CA and their carers 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.

### **Disclaimer**

This leaflet is for information purposes only and, while every care is taken to ensure its accuracy, no guarantee of accuracy can be given. Individual medical advice should be sought before taking or refraining from taking any action based on the information contained in this leaflet and nothing should be construed as medical advice given by Ataxia UK or any of its officers, trustees or employees. No person shall have any claim of any nature whatsoever arising out of or in connection with the contents of this leaflet against Ataxia UK or any of its officers, trustees or employees.

This leaflet was produced by Ataxia UK.

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As soon as you contact us we create a record in your name.

Information we collect, including information you give us during your first enquiry and any subsequent correspondence with us, is added to that record. Your personal data are used to provide you with the services, products or information you have requested and to further our aims as a charity.

The type of information we collect might include name, address, telephone number, email address and bank details. We may also collect "sensitive" data such as ethnic origin, physical or mental health, sexuality or criminal offences. All your personal data are held securely and transfers of data within Ataxia UK are on a "need to know" basis only.

If you provide us with personal data, including sensitive data, you consent to us using that data for the purposes for which it is intended.

If you have any questions regarding data protection, or would like to receive a copy of our Privacy Statement, please contact Ataxia UK. 

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This leaflet has been awarded a Triangle mark by the Centre for Health Information Quality, based on accuracy, clarity and relevance.

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