

## ATAXIA: WHAT'S THAT?

The European Reference Network – Rare Neurological Diseases wish to gratefully acknowledge the significant input from Ataxia UK for this general leaflet on Ataxia.

EUROPEAN REFERENCE NETWORKS  
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES

**Share. Care. Cure.**



# ataxia: what's that?

There are many different types of ataxia that affect people in a variety of ways. This booklet gives a general introduction to ataxia and what to expect when you are being investigated if you have ataxia.

## Disclaimer:

We have made every effort to ensure that the information in this booklet is up-to-date, unbiased and accurate. We hope that this will complement any professional advice you receive. Please do continue to talk to your health and social care team.

The leaflet was adopted for use in the ERN by the patient advocate Dr Mary Kearney, Ireland. The medical sections of this booklet were originally written by ataxia- expert neurologists, Dr Paola Giunti (National Hospital for Neurology and Neurosurgery, London) and Dr Rajith de Silva (Queen's Hospital, Romford, London) for Ataxia UK. The information has been reviewed and adapted for European dissemination by Drs Caterina Mariotti & Sylvia Boesch members of the European Reference Network for Rare Neurological Diseases in August 2020 as well as endorsed by the Cerebellar Ataxias & Hereditary Spastic Paraplegias Disease Group of ERN-RND.

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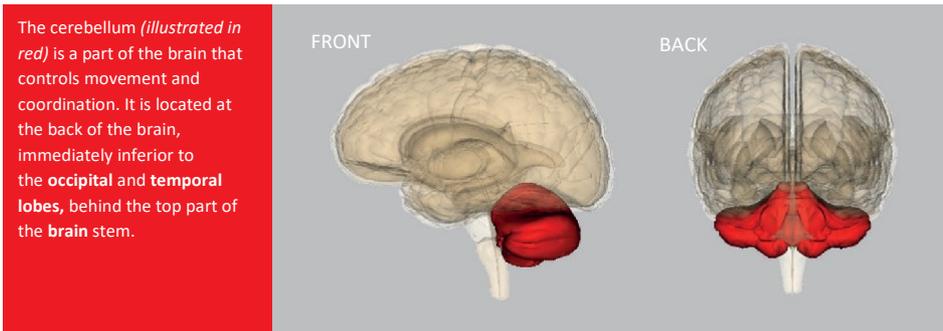
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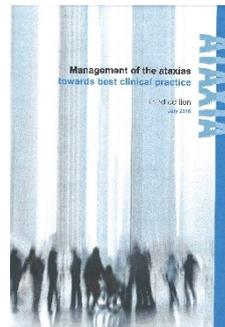
## WHAT IS ATAXIA ?

Ataxia is a symptom, it is not a diagnosis. Ataxia means ‘lack of order’ and is used by doctors to describe problems with balance and coordination. The conditions covered in this booklet are mostly those in which the ataxia is permanent and, in many cases, progressive (ie. the symptoms get worse with time).

Many types of ataxia are described as **cerebellar ataxias**. ‘Cerebellar’ means anything to do with the cerebellum, a part of the brain controlling movement and coordination. There are many different types of cerebellar ataxia: some types are not hereditary, whereas a large number of ataxias are hereditary. Some types have been found in only a few families in specific countries, while others are more common and are found throughout the world. There are no accurate figures for the prevalence of ataxia in Europe but it is considered that well over 25,000 people with ataxia in Europe, so even though it is rare, it is not as rare as you might think.



You may find that many people, even some doctors, have never heard of specific types of ataxia, so it could be helpful to give them a copy of this booklet. You could also give your doctor a copy of Ataxia UK’s guide for health professionals which is called *Management of the ataxias: towards best clinical practice* (or the summary for GPs). Both publications are available free of charge from Ataxia UK ([www.ataxia.org.uk](http://www.ataxia.org.uk))



## Who gets ataxia?

Ataxia can affect anyone at any age, depending on the cause.

## What causes cerebellar ataxia?

There are several causes:

- 1) Inheritance - Some people inherit ataxia via specific genes that have come from one or both parents. Some people may carry a genetic defect causing ataxia which is not inherited from their parents.
- 2) High level of alcohol or prolonged exposure to alcohol
- 3) Damage to the brain, for example from a stroke, tumour, head injury, viral infection or autoimmune disease
- 4) Very occasionally ataxia is due to vitamin deficiencies
- 5) Unknown (idiopathic) - Sometimes it is not possible to find the cause of ataxia despite having had many tests
- 6) Cerebellar malformations

## What symptoms are experienced by people with ataxia?

People with ataxia have problems with coordination and balance. Often people first notice a problem when they realize they have been falling over more than usual, walking in the dark, struggling to walk in a straight line or have become clumsier than you would expect. As the condition progresses walking may become difficult or even impossible, so people may need to use a wheelchair to get about some or all the time.

Other common symptoms experienced by people with ataxia include:

- Hand clumsiness
- Slurred speech (also called **dysarthria**)
- Problems with swallowing which can cause choking or coughing
- Tremors or shaking, often of the hands
- Fatigue or tiredness
- Problems with sight, or blurred or jumpy vision due to difficulty controlling eye movements
- Bladder problems (i.e. urinary urgency and incontinence)

Specific types of ataxia may also cause other symptoms, for example, in Friedreich's ataxia, the most common ataxia worldwide, it can sometimes be associated with heart problems (**cardiomyopathy**), **diabetes** or curvature of the spine (**scoliosis**).

In most people with ataxia, the ability to think and understand is not affected. However, there are emotional aspects of coming to terms with ataxia and these can vary from person to person.

People with ataxia can experience mood disorders, such as depression, which are treatable. Some specific types of ataxia do affect mental function, but these are rarer forms.

Ataxia affects people in different ways. Some people are affected very mildly, for example they only experience slight balance problems and may walk using a stick. Other people experience symptoms more severely and require assistance from caregivers to carry out everyday living tasks.

Although ataxia can affect people significantly, many people with the condition lead full and active lives, attending school, further education, and training, working, bringing up families and travelling the world.

## Does ataxia change with time?

Most types of ataxia covered in this booklet are known as **progressive**, which means they gradually get worse with time. How quickly this happens depends on the type and cause of ataxia, and on individual factors too. Ataxia usually progresses slowly, with changes taking place over many years, although this does depend on the person. Everyone experiences ataxia differently, and this includes their symptoms.

In some types of ataxia that are inherited, people carry the gene for ataxia, but do not develop symptoms for many years. As a very rough guide, the earlier the ataxia starts, generally the faster it progresses. But there are also early onset and slowly progressive forms. It is not possible to predict what will happen in a particular case. We need more research to find the answers to these questions.

Some types of ataxia are not progressive. For example, disorders that involve malformations of the cerebellum that occurred before birth are usually non-progressive. When children get ataxia as a result of viruses, such as chickenpox, a full recovery generally occurs within a few months. People who get ataxia as a result of a stroke or multiple sclerosis can also make an almost full recovery from the ataxia symptoms.

Ataxia due to traumatic brain injury is usually non- progressive. However, patients with ataxia due to brain tumors may either have a progressive condition or a non-progressive condition (once the tumour has been removed).

## How is ataxia diagnosed?

It is sometimes difficult to get a diagnosis of the specific type of ataxia, as many different conditions can seem very similar. A neurologist may need to do extensive tests to find out exactly what a patient has, and this can take time. Investigations include:

**Patient history:** This usually involves your doctor asking you questions to help identify whether the ataxia is inherited from a family member, if the ataxia is progressing (within weeks to months) your doctor might like to see if it is caused, for example, by a tumour or alcohol poisoning.

**Blood tests:** In the absence of any family history of ataxia, your General Practitioner or specialist will probably do routine blood tests first. The blood tests would usually include:

- Full blood count & C-reactive protein
- Kidney, liver, bone, & thyroid function tests
- Blood sugar,
- Vitamin B12 and folate level
- Vitamin E
- In children: Alpha Fetoprotein



**Family history:** This helps to determine whether the patient has an inherited type of ataxia. If their parents and grandparents also have/had ataxia then it is likely that the patient has an autosomal dominant inherited ataxia (see page 13). If the parents are not (or were not) affected but more than one child has ataxia, it suggests that the condition is inherited recessively (see page 17). Even if no one else in the family is affected, this does not necessarily mean that the ataxia is not inherited.

### Referral to neurologist:

Your general practitioner will most likely refer you for further investigation to a neurologist for further assessment.



**Brain scans:** The neurologist will probably organize a magnetic resonance imaging (MRI) brain scan, which will give an image of the cerebellum and other parts of the brain and show whether they are damaged.

The scans can sometimes be used to rule out more or less common treatable causes of ataxia (tumor, multiple sclerosis, leukodystrophy etc.)

**Genetic tests:** When you have seen your neurologist, depending on the results of examining the person, he/she may decide that the person may have a genetic ataxia and organize a blood sample to verify this. If the result is positive, then it can generally be considered a definite diagnosis.

If the results of the genetic test are negative, this may mean that:

- 1) it is a type of inherited ataxia for which the gene/mutation has not been tested or yet known

2) the ataxia is not inherited.

In these circumstances, with your permission, your blood sample may be stored for a long time so that if there are advances in research and new tests become available, further tests can be done.

**Other laboratory tests:** Your neurologist may do more blood tests especially looking for metabolic or autoimmune causes of ataxia (such as vitamin E deficiency, Wilson disease caused by abnormal copper metabolism, markers of autoimmune conditions, gluten allergy investigation, etc.).

## Pre-symptomatic genetic testing

If you are a close relative of someone with a known inherited ataxia and you do not show any signs of ataxia, it is possible to have a genetic test for yourself. The chance of developing ataxia or being a carrier depends on how the ataxia is inherited (as described above in *family history*).

The decision to have a test or not is a very personal one and can be a difficult one to make. Some people prefer to have all possible information in advance, in order to plan for the future. Others may prefer not to know unless there is a cure. Test results can have a range of long-term consequences, affecting everything from whether to have a family to the ability to get insurance. Support in making this decision is available from clinical geneticists or neurologists, who are experienced in talking to people about these issues.

Tests are only generally available for adults (i.e. people over 18 years old), but this may vary depending on individual circumstances. For more specific advice talk to your doctor.

## Genetic counselling

Genetic counselling is for people at risk to suffer from a serious inherited disorder. The results of a genetic test can bring up some difficult questions and concerns about the future, so it is often useful to see a genetic counsellor or clinical geneticist or experienced neurologist before testing goes ahead, to talk about what the potential results might mean.

Implications of a genetic result involve prognosis for specific symptoms, evolution of the disease, and possible complications. Positive genetic test has also implication for family members and for future generations. If the neurologist diagnoses an inherited condition, they will make a referral for genetic counselling. This provides an opportunity to discuss what a diagnosis could mean for the person with ataxia and their family.

A doctor or neurologist can arrange a referral to a regional genetic centre (where genetic services are available) to explain the tests and the implications of the test results. The availability of this service varies in every European country.

## Are there any treatments for ataxia?

Some of the very rare ataxias are treatable (vitamin E and CoQ10 deficiency, gluten ataxia, or episodic ataxias, for example), which makes it so important for people to get a specific diagnosis of the type of ataxia they have if possible. All of those with ataxia can benefit from a multidisciplinary management approach to help them minimize the associated spectrum of complications that may occur and so that they can adapt to life with ataxia and live life to the fullest.



*International ataxia meeting*

When a family first receives the diagnosis of progressive ataxia, they usually have not heard of the condition or come across other people with it. Support from patient organizations can therefore be particularly important at this time.

The possibility of meeting others in the same situation, receiving emotional support and information, tips on employment, exercise, equipment and house adaptations is invaluable. There is usually an opportunity to learn of research developments (as well as taking part in research projects) via these organizations.

There is a European organization for ataxia called Euro-ataxia ([www.euro-ataxia.org](http://www.euro-ataxia.org)). A lot of European countries have their national patient organizations for ataxia. In fact, many countries have two ataxia organizations – one for Friedreich's Ataxia and the 2<sup>nd</sup> ataxia organization is for all the other ataxias.

### **European Patient Organizations for Ataxias**

Belgium	<a href="#">Spierziekten Vlaanderen</a>
Denmark	<a href="#">Foreningen for Ataksi HSP</a>
Finland	<a href="#">Neuroliitto</a>
France	<a href="#">BRAIN-TEAM list of ataxia patient organisations</a>
Germany	<a href="#">Deutsche Heredo-Ataxie Gesellschaft (DHAG)</a>
Ireland	<a href="#">Ataxia Foundation Ireland</a>
Italy	<a href="#">Associazione Italiana per la lotta alle Sindromi Atassiche</a>
Netherlands	<a href="#">ADCA/ataxie vereniging</a>
Norway	<a href="#">Norwegian Association for Hereditary Spastic Paraplegia/ Ataxia</a>
Poland	<a href="#">Polish Association for Families with Spinocerebellar Ataxia (Forum Ataksja)</a>
Spain	<a href="#">Federación de Ataxias de Espana (FEDAES)</a> <a href="#">Asociacion Catalana de Ataxias Hereditarias (ACAH)</a>
Switzerland	<a href="#">Schweizerische Muskelgesellschaft</a>
UK	<a href="#">AtaxiaUK</a> , <a href="#">Ataxia Telangiectasia Society</a>

### **Patient organizations specifically for Friedreich's ataxias**

Australia	<a href="#">FARA Australasia</a>
Belgium	<a href="#">Association Belge de l'Ataxie de Friedreich (ABAF)</a>
France	<a href="#">L'Association Française de l'Ataxie de Friedreich</a>
Germany	<a href="#">Friedreich Ataxie Förderverein e.V.</a>
Ireland	<a href="#">FARA Ireland</a>
Italy	<a href="#">GoFAR</a>
Sweden	<a href="#">Bota FA! Sweden</a>
Switzerland	<a href="#">Association Suisse de l'Ataxie de Friedreich</a>
USA	<a href="#">Friedreich's Ataxia Research Alliance (FARA)</a>

### **Patient organizations specifically for Dominant Ataxias**

Israel	<a href="#">The Israeli Machado Joseph Association (SCA 3)</a>
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## What about a cure?

*“Hearing about the latest research news always fills me with hope that we will one day have a cure.”*

At the moment there is no known cure for most types of ataxia. However, there are many clinical trials ongoing and these may result in treatments. In particular, there are many trials taking place testing medications for Friedreich’s ataxia.



*Prof. Ludger Schöls, ERN-RND clinical coordinator  
& neurologist, University Hospital Tübingen, Germany*

In addition, although there may not be a cure, there are many ways to help people manage some of the symptoms they experience (see page 18 onwards in this booklet for advice on living with ataxia).

A lot of progress is being made in finding new genes causing ataxia, which will result in more people getting a specific diagnosis. Many promising new treatments are being tested either in animal models of ataxia or in human trials and may in the future be available to patients.

## TYPES OF ATAXIA

Some types of ataxia are inherited (meaning they are caused by genes passed on by parents to their children) and some are not. When ataxia is not inherited there can be a number of different causes. The various types of ataxia are explained over the following pages.

### Inherited ataxia

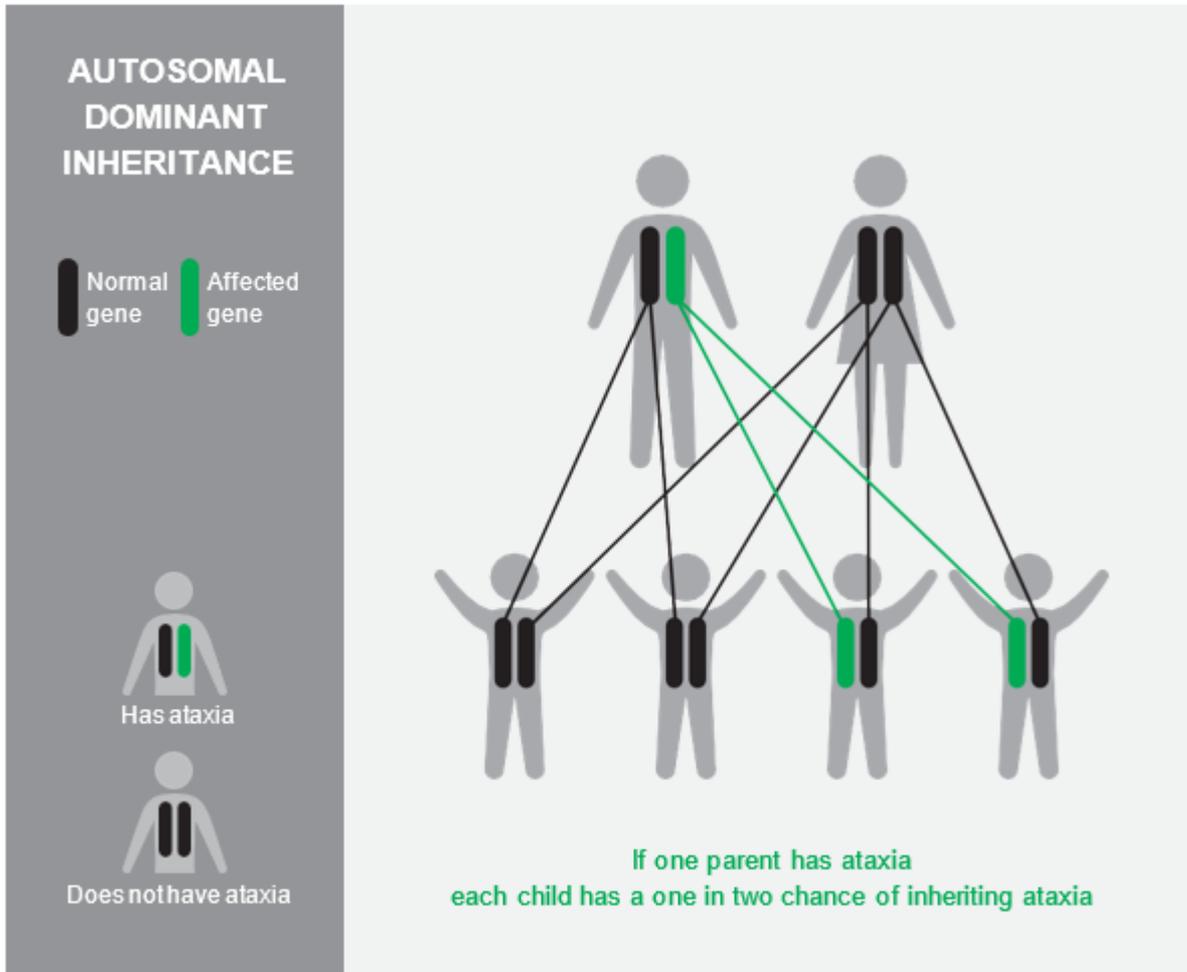
The inherited types of ataxia involve a fault in a gene or genes, which may then be passed on through the generations. They can be divided into four groups, depending on how they are inherited. These are:

- **Autosomal dominant:** meaning that the condition develops after a faulty gene is inherited from one parent only
- **Autosomal recessive:** meaning the condition is only passed on by receiving the faulty gene from both parents. For most genes, each person inherits two copies of the gene: one from their mother and the second copy from their father.
- **Mitochondrial:** meaning you inherit ataxia from the maternal (mother's) line
- **X-linked:** in rare cases, ataxia can result from faulty genes that reside in the X chromosome, and in these cases either just men are affected, or men are more severely affected than women.

## Autosomal dominant inheritance

In this case, ataxia is caused by having one copy of the faulty gene, inherited from only one parent. In ataxia of this kind there is a one in two chance of passing the ataxia on to each child. A genetic counsellor or clinical geneticist can explain this further and discuss the implications of having children.

In some types of inherited cerebellar ataxia of autosomal dominant type, the condition becomes more severe as it gets passed down the generations, and the age of symptom onset gets younger. This is called **anticipation**.



Autosomal dominant ataxias continued:

## Spinocerebellar ataxia

Several spinocerebellar ataxias (also called SCAs) have been identified, all of which are caused by faults in different genes. As each gene is found, it is given a number; for example, SCA1, SCA2, SCA3 and so on. Although each type is caused by a different gene, the SCAs are often very alike and sometimes it is only possible to tell the difference between them by doing genetic tests.

At present, we know of more than 50 different SCAs. The prevalence varies greatly depending on the country. Some of the subtypes have been found only in few families all over the world, others are more common. Routine genetic tests are not yet available for all of them. Specific tests are available for 21 SCAs, but only some of these are available routinely. Tests include: SCAs 1, 2, 3, 6, 7, 12, and 17. In some cases, depending on individual factors and ethnic group, i.e. dentatorubral-pallidoluysian atrophy (DRPLA) testing is now available.

A new technique known as ‘next-generation sequencing’ (NGS) may make testing for a wider range of ataxias. In particular it makes screening for SCAs more accessible. Although NGS has an enormous diagnostic power, data interpretation remains challenging because of the high incidence of novel and ultra-rare benign variation on genes and false association of genes with disease in the literature. In addition, NGS cannot be used for all ataxia subtypes as it usually does not capture SCA caused by repeat expansions.

Further information can also be found in [Management of the Ataxias: towards best clinical practice](#) for medical professionals produced by Ataxia UK.

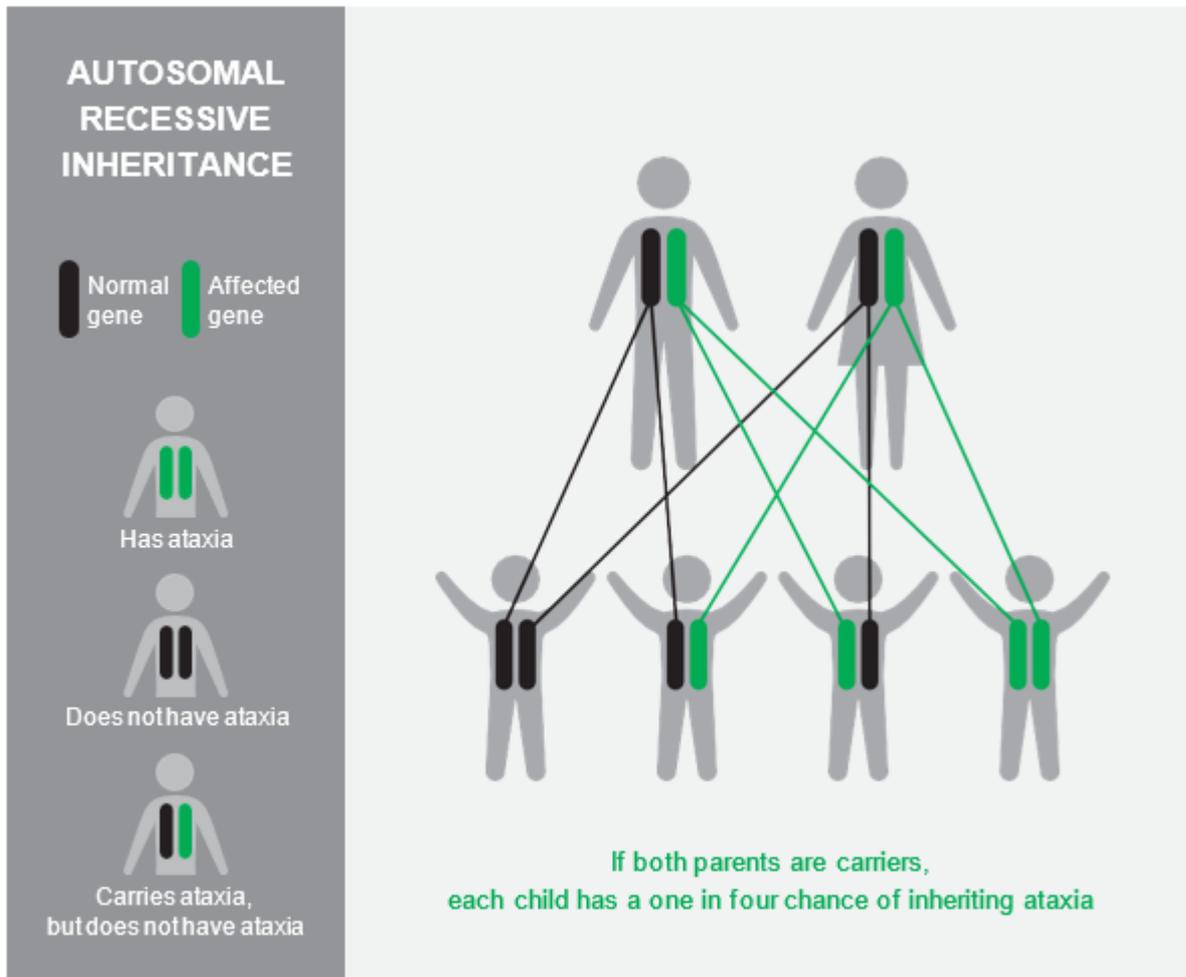
## Episodic ataxia type 1 (EA-1)

EA-1 is different from most other types of ataxia because it involves short attacks in which people lose coordination and may slur their speech, generally for several minutes. EA-1 is usually not progressive, meaning it does not tend to get worse, except in some older people. Attacks sometimes happen spontaneously with no obvious cause, they can be triggered by a sudden shock or movement or by being tired, anxious, or stressed. Treatment with some epileptic drugs (i.e. carbamazepine) may lessen the attacks and reduce their intensity.

## Episodic ataxia type 2 (EA-2)

In EA-2, attacks of ataxia can last for hours or even days. Treatment with **acetazolamide or aminopyridines** may prevent or lessen the attacks (or episodes) but the use of any medicines must always be discussed with a doctor. As stress often triggers attacks, stress management techniques may also help. EA-2 is caused by a mutation affecting the same gene as in SCA6 (which has a different type of mutation). This is also the gene involved in a form of inherited migraine, called familial hemiplegic migraine. There can be some progression in the symptoms of EA-2 over time.

## Autosomal recessive inheritance



In these cases, ataxia is caused by having two copies of a faulty gene, one inherited from each parent. In other words, a child can be born with ataxia of this kind if both parents have one faulty copy of the gene. This means the parents are **carriers** of ataxia, although they do not have ataxia themselves.

If two parents are carriers there is a one in four chance of them having a child with ataxia, and there is a one in two chance of having a child who does not have ataxia but who also carries the faulty gene. If the child is a carrier, they may pass it on to their own children. There is also a one in four chance that a child will neither have ataxia nor be a carrier.

In this situation a clinical geneticist can advise on how these genes are inherited and the implications for other family members.

There are over 30 types of ataxia inherited in an autosomal recessive way, some are well known including Friedreich's ataxia. Other ataxias are less common, and a few autosomal recessive ataxias affect only a few families worldwide.

## Friedreich's ataxia

Friedreich's ataxia (FA) is the most common type of inherited ataxia worldwide, mostly affecting children and teenagers; on average, the symptoms start between 5 and 15 years of age. It initially causes clumsiness of movement and progresses to unsteadiness in standing and walking with wheelchair dependency by late teens or early twenties. Speech usually becomes slurred. Other major problems which can develop include a curved spine (scoliosis) foot deformity (a high arch), diabetes mellitus and heart problems which are the cause of death in 60% of those with Friedreich's ataxia.

## Ataxia-telangiectasia

The first signs of this ataxia usually appear early in childhood, when the child is beginning to walk they wobble and sway. Somewhat later, they often develop problems moving their eyes. After some years, they may develop 'telangiectasia' tiny red spider veins at the corner of the eyes, surface of the ears and cheeks. Later they may develop immune system problems which may lead to recurrent respiratory tract infections and a predisposition to cancer.

There is an international patient support group called 'AT Children's Project'. **Website:** [www.atcp.org](http://www.atcp.org)

## Other autosomal recessive ataxias

- Ataxia with oculomotor apraxia types 1 or 2 (known as **AOA1** and **AOA2**)
- Ataxia with familial isolated vitamin E deficiency AND Abetalipoproteinemia
- Cerebellar ataxia with muscle coenzyme Q<sub>10</sub> deficiency
- Early-onset cerebellar ataxia with retained tendon reflexes
- Infantile onset spinocerebellar ataxia
- Marinesco-Sjogren syndrome
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)
- Joubert syndrome
- Non progressive congenital ataxias with shrunken cerebellum (AD , X-L or AR traits)
- Non progressive congenital ataxias with cerebellar hypoplasia (global or vermis) (AD, AD , X-L or AR traits)

It is quite possible in the years to come that more information will become available about these ataxias and more ataxias will be discovered.

## Mitochondrial conditions

These types of ataxia involve changes (or **mutations**) in the genes that code for proteins in mitochondria, the energy-producing compartments of cells. As each person inherits their mitochondria and mitochondrial genes from their mother, this type of condition can only be passed down the maternal line, i.e. from the mother. Women who have such a condition are at risk of passing it on to their children (male or female).

Most of the genes found in the mitochondria are involved in the production of energy, so generally mitochondrial disorders are caused because cells cannot produce enough energy, which prevents them from carrying out their normal functions. As muscles and the brain need a lot of energy to function, they are the most likely parts of the body to be affected by mitochondrial disorders. Some mitochondrial disorders have ataxia as a main symptom.

Examples of mitochondrial ataxia conditions are:

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

## X-linked inherited ataxias

Every cell in the body has 23 pairs of chromosomes - long stretches of DNA containing many genes. In humans, gender is decided by one of these pairs, known as the X and Y chromosomes. While women have two X chromosomes, men have one X and one Y. This can mean that some conditions with faulty genes on the X chromosome are more likely to affect men (and on the rare occasions that females are affected, it is generally much more mildly than in men). Females can be carriers of a faulty X chromosome gene and pass on a condition to their sons. Haemophilia is an example of a condition that is inherited in this way, and some forms of ataxia can also be X-linked.

## Non-inherited cerebellar ataxias

Some people who have ataxia have no history of ataxia in the family. However, they may still have a type of ataxia that can be passed on to their siblings. It could be that they are the first member of the family to have developed a mutation in a gene causing an inherited ataxia or their parents may have passed away before developing signs of ataxia. Alternatively, they may have a non-inherited form of ataxia.

If the ataxia is not inherited it is sometimes called **sporadic cerebellar ataxia**, and if the cause of the ataxia is not known it is sometimes called **idiopathic cerebellar ataxia**. For example, a number of people are diagnosed as having idiopathic late-onset cerebellar ataxia, which means that the condition occurs later on in life and its cause is unknown. People can be diagnosed with this if there is lack of evidence of a genetic or other cause. It often progresses slowly and has few additional symptoms.

Examples of non-inherited ataxia include:

### 1) Multiple system atrophy with cerebellar features (MSA-C)

This is a condition that occurs later in life. It is a progressive cerebellar ataxia and has its own support charity.

In UK - Multiple System Atrophy Trust [www.msatrust.org.uk](http://www.msatrust.org.uk) or the USA patient organizations: [www.mutpilesteyemstrophy.org](http://www.mutpilesteyemstrophy.org) or [www.brainsupportnetwork.org](http://www.brainsupportnetwork.org). There is no European organization for MSA-C at present.

## Undiagnosed Ataxia

Knowing that something is wrong with you or your child and you have not even got a specific diagnosis, it leaves everyone in the difficult position for several reasons. In the case of a child, knowing that your child is different from others but not knowing why or what to do about it, it can be very difficult. The search for a diagnosis can be anguishing for both children and parents. Such people often join the ataxia organization in their own country even in the absence of a specific diagnosis.

There are several 'Facebook' groups for rare charities, and it is not uncommon for patients to get support from such groups. However, one should always be wary that there are people and companies only too willing to take money from unsuspecting people who have an untreatable condition.

The [SWAN \(Syndromes With a Name\) UK](http://www.swan-uk.org) is a non-profit, self-help organization and a registered charity which promotes awareness of challenges faced by children and families affected by undiagnosed diseases. It campaigns for equal rights and recognition, is building a database to help with future research, links families to others when possible, and facilitates the exchange of information and stories through its newsletter.

## LIVING WITH ATAXIA

In this section you will find information on the practical aspects of day-to-day living with ataxia. There are many different ways to improve quality of life when living with ataxia.

### What can help in living with ataxia?

Although there is currently no cure for ataxia, there are a number of treatments available to help with the symptoms people experience. Medications are available, for example, for muscle spasms, tremors, bladder problems, abnormal eye movements and depression. Cardiac problems seen in Friedreich's ataxia are also treatable.

It is usually recommended that people with progressive ataxia be seen regularly by a neurologist (at least annually) who can monitor the condition and provide help with any new problems that may have emerged. It also gives you a chance to benefit from any new medical advances.

Physiotherapy and exercise such as swimming, weight lifting, cycling, horse-riding may prevent loss of strength, preserve mobility and help with transfers for those who need to use a wheelchair.



Speech and language therapy can help with problems involving speaking, swallowing, coughing, choking and, if needed, communication aids, such as some computer programs.

Occupational therapy is also important; for example, with home adaptations, teaching strategies for daily activities or when buying a wheelchair.

Meeting other people with ataxia is well known to help those with ataxia as they realize they are not alone in what they are going through this illness. Patient organizations, national and international online forums are particularly helpful in this regard. People usually communicate through English on international forums.

### Your rights

The European Disability Forum (EDF) was created in 1996 and is an umbrella organization of persons with disabilities that defends the interests of over 100 million persons with disabilities in Europe.

It helps ensure that decisions at a European level concerning people with disabilities are taken with and by persons with disabilities. The vision of EDF is that people with disabilities in Europe are fully included in society on an equal basis with others and that our human rights as outlined in the United Nations Convention the Rights of persons with Disabilities (UN CRPD) are fully respected, protected and fulfilled.

The [United Nations Convention on the Rights of Persons with Disabilities \(UN CRPD\)](#) is an international human rights treaty that reaffirms that all persons with disabilities must enjoy all human rights and fundamental freedoms. It clarifies that all persons with disabilities have the right to participate in civil, political, economic, social and cultural life of the community just as anyone else. The CRPD clearly stipulates what public and private authorities must do to ensure and promote the full enjoyment of these rights by all people with disabilities.

It is unlawful to discriminate against disabled people in various areas of their lives, including work, education, travel and leisure. Sex discrimination and race relations are also covered by the Equality Act.

## Counselling and emotional support

People with ataxia often need to talk to a counsellor or therapist to discuss some of the issues ataxia has brought up for them. The researchers and doctors at the European Reference Network for Rare Neurological Diseases are very conscious of the affect that ataxia has on the activities of daily living. It is now mandatory in all clinical trials, in addition to the primary outcome which is usually cardiological or neurological to include an outcome with respect to the effect the intervention has on the daily life activities of those with ataxia.

## Being a caregiver

Increasingly, more support is available for carers looking after a person with ataxia. Traditionally family members or loved ones have often been the sole caregiver for the person with disability. It is generally considered that is better not to have a relative as the primary caregiver for the person with ataxia if circumstances allow. The economic circumstances of each European country will possibly dictate how much 'outside' help a person with ataxia may qualify for.

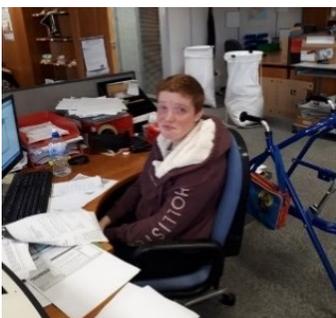
Where family members do care for the person with ataxia, it is vital for them to take time for themselves so that they are refreshed and rested, and their own health does not suffer. Many caregivers of people with ataxia find it very helpful to come to patient organization meetings and events to get some support for themselves from others in similar circumstances.



## Education

Although schools vary as to how accessible they are, in general, modern facilities can accommodate pupils with physical impairments, and improvements are continually being made to accessibility. Such information can often be found in a school's accessibility plan, a copy of which must be given on request.

Disabled students ideally need help to buy specialist equipment e.g. laptop, a note-taker and extra travel costs required.



## Employment

Many people with ataxia continue to work after their diagnosis and hold down jobs for many years. There are usually a number of schemes to help with this, but it depends on the country. Usually, the ataxia patient organization may be able to help you source information.

## Housing adaptations

Some people need to make adjustments to their home when they develop ataxia. An occupational therapist with social services can advise on the home adaptations required. It is not uncommon for local authorities to give different kinds of grants to private sector owner-occupiers and public housing developments tenants. These include grants to make properties fit to live in and to cover improvements and adaptations.



*Dad who had ataxia with his family*

## Planning a family

Many people with inherited types of ataxia have children. Some people develop ataxia after they have had children, and others may do so while knowing that they have ataxia. Every person with ataxia will have a different view on whether to have children. It is always a deeply personal decision.

If someone has a known recessively inherited ataxia (e.g. Friedreich's ataxia) and they wish to start a family, their partner can be tested to see if they are likely to be a carrier of the same type of ataxia. If he/she is not a carrier, then their child is highly unlikely to develop that type of ataxia.

## Walking aid

When considering the use of a walking aid, it is best to consult an occupational therapist for help and advice if they are available to you. Disabled living centres offer a range of equipment as well as advice and information but often one will get ideas about walking equipment at a national patient organization meeting.

In general, children find walking sticks tend to trip them up. Walking frames are usually more suitable for those who have some ability to stand and walk but need help keeping their balance. A walking frame offers more stability and support, and many have additional features such as wheels, brakes or a seat for resting.

Other forms of mobility aids include gait trainers, which have a frame and provide more support than a standard walker, and scooters, which may be used as a form of transport over a longer distance.



*This is a posterior walker with extra arm support*



*Helen has used her wheelchair for long distances since she was 20 years old.*

*Note the 'widget' device in her left hand which is attached to both wheels. Its use propels the chair and prevents her having to replace her hands on the wheel after each turn.*



*Al uses an electric chair. He had ataxia symptoms since he was 11 years old. He was diagnosed 10 later with Friedreich's Ataxia. He recently completed a Degree in Disability studies having previously studied Design. He is now 54 years old.*

## Manual and electric wheelchairs

Although not everyone with ataxia uses a wheelchair, many people find it makes life easier. Some people can walk short distances or stand for a short period; they may use a wheelchair for the rest of the time.

As a parent, you might get upset thinking about your child having to use a wheelchair. However, as the ataxia progresses, you will soon realize that if your child uses a wheelchair it actually gives them greater independence and preserves their energy for important things.

Diversity in manual and electric wheelchairs is growing all the time. Technological developments mean chairs can be made stronger, faster, and lighter than ever before. There are many different types of manual and electric wheelchairs, including sports wheelchairs, standing wheelchairs and transport wheelchairs.

Factors such as age, need and ability are all important when finding a suitable wheelchair. The cost can vary from hundreds to tens of thousands of pounds, depending on what the chair is made of and whether it has been made to measure.

Manual wheelchairs have the advantage in that they are:

- 1) easier to transport than electric chairs
- 2) help the person with ataxia to keep themselves fit

They have the disadvantage that the repeated action of using the wheels can cause recurrent shoulder problems which is where the widget or similar device could help.

The main advantage of using an electric wheelchair rather than a manual wheelchair is that it is less physically demanding. All operating is done by battery, and you do not need to rely on assistance to move.

*"At first I felt embarrassed being in a wheelchair, but my friends and family were a great source of strength."*

## Assistance dogs



*“My dog is amazing - he can even help me empty the washing machine! He really helps me to live independently.”*

Assistance dogs are specially trained to help disabled people carry out everyday tasks that they would otherwise find difficult. People with ataxia sometimes find that having an assistance dog helps them to retain their independence in addition to the dog being a wonderful companion to have. Dogs can be trained to carry out a variety of tasks in order to help people live as comfortably as possible. Tasks can include opening and closing doors, reaching up to shop counters, carrying a shopping basket or even emptying the washing machine.

## On the road



*The driver is entering the car from the rear of the vehicle using the wheelchair.*

Learning to drive is a great way to get mobile. The minimum age for learning to drive depends on your national authority. The earlier the person who has the ataxia learns to drive the better chance they have of being successful. It helps to give them a little independence at a sensitive age in their life.

Many people with ataxia may be eligible for parking concessions for drivers who have ataxia or passengers who have problems walking. The concessions allow you park close to your destination. To apply for this concession, contact the social services department of your national or local authority.



If you drive, most European countries would expect you to inform your National Driver authority as soon as you are diagnosed with ataxia. This does not automatically mean you have to stop driving. For some people with ataxia, their condition means they need adaptations to their car to carry on driving and eventually may decide to give up driving. On the positive side, you may well qualify for concessions which can facilitate parking close to venues.



## Getting out and about

Most cinemas, especially multiplexes, offer good facilities for people with ataxia and other impairments. Details of these can usually be found on their websites. In recent years much public transport and regular taxis have been made accessible for people with mobility impairments. Most rail, bus, and airline companies will allow assistance to be booked in advance if you need some help to travel, for example to change trains. Contact the individual firm for more information.

Transport in major cities who host the Paralympic game every 4 years make special efforts to have the public transport as accessible as possible during the Paralympic games. It is an ideal time to visit major cities.



## Sport and leisure

Many sports can be adapted to allow people with conditions like ataxia to take part. This is reflected by the increase in the range of sports at the Paralympics. If you enjoy watching sport, many sporting venues accommodate wheelchair users at reduced rates.



## Getting online

Many people with ataxia find that going online can be a great way to keep in touch, arrange travel and get their shopping done. Most websites and internet browsers have accessibility options that allow the font size to be changed or show a text-only view, and so on. A keyboard and mouse can be adapted for ease of control and there are also various communication aids that can help with computer use, such as voice recognition software. The computer desktop and settings can also be customized to make them more accessible. Mobile phones can also be adapted for easier use, and some (such as the iPhone) offer voice recognition apps at a fraction of the price.



## Holidays and travel

Most holiday services have accommodation for people with impaired mobility and can offer extra facilities for those with additional needs. This is yet another place where the patient organization may be able to help you. While many of older areas in the bigger cities in Europe may not have been built for those with wheelchairs, it is possible to get around with a little advance planning. Most cities provide a guide to help those with mobility difficulties.

## What's next?

Although there is no hiding the fact that ataxia does affect people, it does not have to prevent people from having a full, active, and enjoyable life. We all live in hope of new treatments.

**We hope you have found this booklet useful. Your feedback is always welcome. Help us improve the next edition by telling us what you think: [info@ern-rnd.eu](mailto:info@ern-rnd.eu)**

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for rare or low prevalence complex diseases

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