

This document is intended to provide patients, families, and their caregivers with basic information regarding how physicians classify the different forms of Ataxia. Correctly defining the type of Ataxia is an important step in a patient's evaluation and can help guide their medical workup, suggest potential treatment strategies, and even indicate if appropriate clinical or research studies are available. Many different terms have been used to describe Ataxia over the years and have fallen out of favor for various reasons. The terms listed here are ones currently used by many physicians who treat patients with Ataxia.

Ataxia is a neurological symptom seen by your physician during a physical examination. The definition of Ataxia is a specific problem with balance and coordination not due to muscular weakness. Ataxia can be caused by problems in the inner ear or nerve damage in the legs, but most often Ataxia describes damage to a part of the brain called the **cerebellum**. Doctors may also refer to this problem as **Cerebellar Ataxia**.

Cerebellar Ataxia is classified by its cause (see Figure 1). Unfortunately, there are many causes and the terminology can become confusing. In general, there are two main classes of Ataxia; acquired and genetic.

Two additional classes of Ataxia, idiopathic and unknown, are also sometimes used. It is not uncommon for patients to undergo detailed medical evaluations before the cause is known and their Ataxia can be properly classified.

Acquired Ataxia has an external cause which means a person developed it because of something that happened during their life. This can include many different problems and events such as vitamin deficiencies, autoimmune conditions, some infections, exposures to toxic substances or drugs (especially alcohol), various cancers, and many, many more. Acquired Ataxias often appear "out of nowhere" and the medical term used for this is **sporadic**.

Genetic Ataxia has an internal cause which means it is due to inherent damage in a person's DNA (their "genetic blueprint"). A person's DNA is made up of 23 pairs of chromosomes (mother and father each donating one pair). These chromosomes contain sets of genes which together code for all the information which makes a person who they are. Every person has 20,000 genes and we now know of many specific examples where damage to a certain gene causes Cerebellar Ataxia. Genetic Ataxias run in families (some physicians may say it is an **hereditary** or **familial** Ataxia when this occurs), but it can also be sporadic without a known family history.

- If an Ataxia passes from generation to generation it is said to be **dominant** and is due to a single copy of a defective gene passing from parent to child. Dominant Ataxias are most often seen in adults and some can get worse with each successive generation. The most well-known dominant Ataxias are the spinocerebellar Ataxias (or SCAs) such as SCA1, SCA2, SCA3, SCA6, and SCA7, which are each caused by defects in specific genes. SCA3 is the most common of these worldwide. Specific genetic testing is available for many of these Ataxias. Because the term "SCA" is so widely associated with dominant genetic Ataxias, it should not be used to describe a patient with an unknown Ataxia.

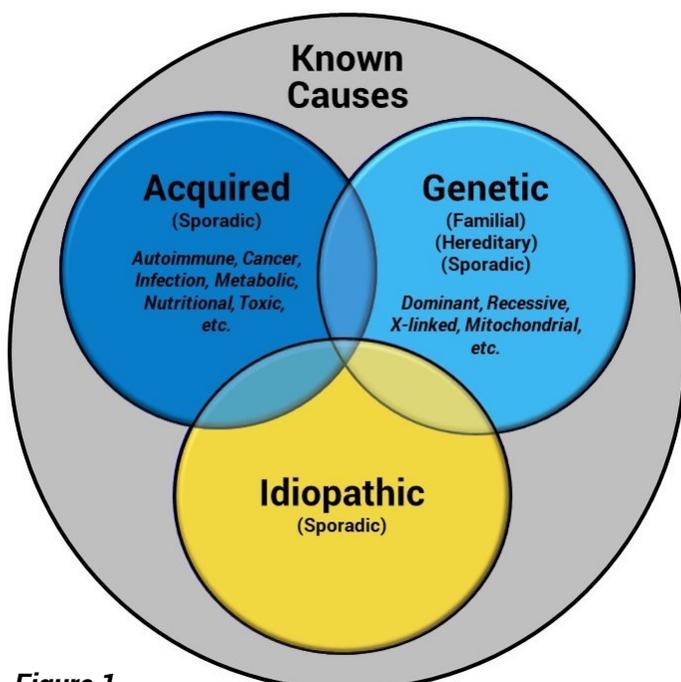


Figure 1

- If an Ataxia is seen in a single generation (e.g., in multiple children but not the parents) it is called **recessive** and is due to two copies of a defective gene; one passed from each parent (who are **carriers**) to the child. Often, recessive Ataxia is seen in children but can also be seen in adults. The most common recessive Ataxia worldwide is Friedreich Ataxia. Specific gene testing is available for many of these diseases as well.
- Ataxia can be inherited in other ways also. **Mitochondrial** Ataxias pass from mother to child through the defective Mitochondria in the mother's eggs. **X-linked** Ataxias are caused by a single defective gene on one X-chromosome, but are often only seen in males because women have two X-chromosomes while men only have one.

As more Ataxia genes are discovered, more and more genetic tests become available. In some cases, it maybe useful to test a patient for problems in a single gene or even a select few genes. Because many genetic Ataxias can look alike (and other rare genetic diseases can sometimes look like a genetic Ataxia), more extensive testing may be needed. Recently it has become possible to examine all the genes in a patient at once (this process is called "whole exome sequencing") and your physician may suggest this. If genetic testing is being suggested, it is always important to understand the type of test being offered and what its potential benefits and shortcomings are to your condition. A genetic counselor should be consulted prior to genetic testing.

Idiopathic Ataxia is a term used when physicians can identify a specific kind of Ataxia but don't yet understand medically or scientifically why a person developed it. Multiple System Atrophy (MSA) is the most common of these. Idiopathic Ataxias are often sporadic. Many scientists and physicians suspect that these types of Ataxia have a combination of internal and external causes meaning certain gene problems and certain life events are acting together, but this is still under investigation.

Unknown Ataxia is a term used when, despite all possible testing, the cause of Ataxia remains undetermined. Unknown Ataxias are usually sporadic, but could also be described as familial Ataxia if the affected gene is not known. Eventually, as physicians learn more about the different kinds of Ataxia, it is hoped that all patients with unknown Ataxias will be definitively assigned into a specific categories.

It is very important for all patients with Ataxia to seek proper medical care. Often this may include a visit to a specialist with expertise in neurology, neurogenetics, medical genetics, or a related field. You may be asked to have an MRI of your brain and various blood tests. Patients with a known or suspected genetic Ataxia may also be asked to meet with a genetic counselor. Patients with all forms of Ataxia could be asked to participate in research so physicians can better understand the disease and learn new information to help others.

The National Ataxia Foundation (NAF) is committed to providing information and education about Ataxia, support groups for those affected by Ataxia, and promoting and funding research to find the cause for the various forms of Ataxia, better treatments, and, hopefully someday, a cure. NAF has been at the forefront funding promising worldwide research to find answers.

Ataxia research has moved into the clinical phase, and pharmaceutical companies have begun recruiting participants for clinical trials. Individuals with Ataxia or who are at-risk for Ataxia are encouraged to enroll in the CoRDS Ataxia Patient Registry. To access the Registry, go to NAF's website www.ataxia.org and click on the "Enroll in the Patient Registry" tab and follow the directions on the CoRDS website.

NAF provides accurate information for you, your family, and your physician about Ataxia. Please visit the NAF website at www.ataxia.org for additional information, including a listing of ataxia support groups, physicians who treat Ataxia, social networks, and more. For questions contact the NAF directly at 763/553-0020 or naf@ataxia.org.

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