

FREQUENTLY ASKED QUESTIONS ABOUT...

Ataxia Classification

Patients often ask the familiar question “What kind of ataxia do I have?”. This document is intended to provide patients, families, and their caregivers with basic information regarding how doctors 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 doctors 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 their cause is known and their ataxia can be properly classified.

Acquired ataxia has an external cause, meaning a person develops it because of something that happens 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**. Unfortunately, ataxias from all other categories can also occur sporadically, so this term is not useful to categorize the cause of the ataxia. Acquired ataxias are important to recognize early because, in some cases, they may be treatable.

Genetic ataxia has an internal cause, meaning 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 (one pair each from their mother and their father) containing sets of genes which together code for all the information which makes a person who they are. People each have over 20,000 genes and we now know of many specific examples where damage to a certain gene causes cerebellar ataxia to develop. Genetic ataxias often run in families (some doctors may say it is a **hereditary** or **familial** ataxia when this occurs) but 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 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

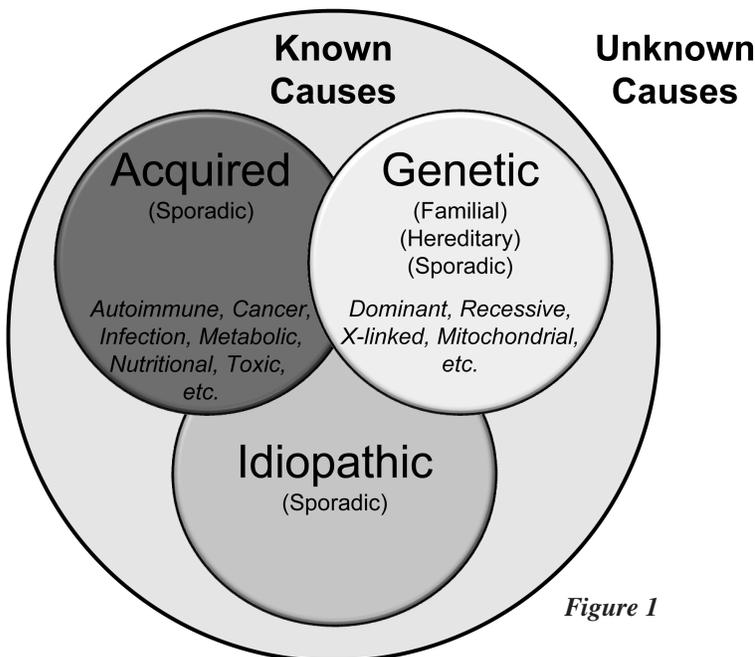


Figure 1

most common of these worldwide. Specific genetic testing is available for many of these diseases. 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 (see below).

- 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 called **carriers**) to the child. Often recessive ataxias are 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.
- In most cases, genetic ataxias are **progressive**, meaning that the symptoms get worse over time. There are also a few forms with recurrent, or **episodic**, ataxia symptoms that often seem to come and go.
- 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 (or are more severe) because women have two X-chromosomes while men only have one.

As more and more ataxia genes are discovered, more and more genetic tests become available. In some cases it may be 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 doctor 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.

Idiopathic ataxia is a term used when doctors 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 describe familial ataxia if the affected gene is not known. Eventually, as doctors learn more about the different kinds of ataxia, it is hoped that all patients with unknown ataxias will be definitively assigned into one of the other 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, neuro-genetics, 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 doctors 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, services to individuals affected with various forms of ataxia, and promoting and funding research to find more causes, better treatments, and, hopefully someday, cures. NAF can help by providing 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, online chat groups, social networks, and more. For additional questions please contact the NAF directly using the information listed below.

The National Ataxia Foundation is grateful to Brent L. Fogel, MD, PHD