

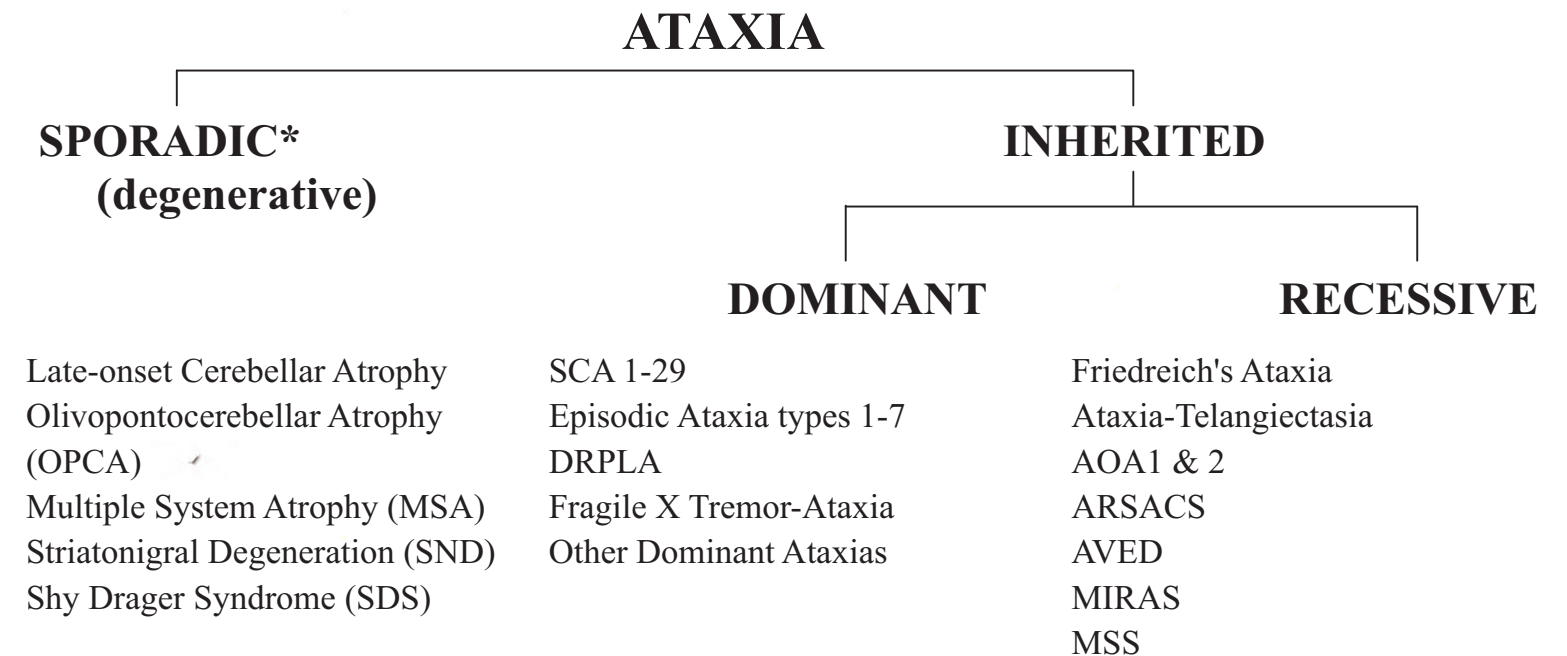
FREQUENTLY ASKED QUESTIONS ABOUT...

CLASSIFICATION OF ATAXIA

Trying to classify a rare neurological disease such as Ataxia is very difficult. It has been tried many times in the past, which accounts for the myriad of names given to different types of ataxia.

With the availability of better brain imaging and molecular genetic testing, classification of ataxia is becoming more defined. It will always be evolving as more knowledge is gained, especially through genetics. The goal of this particular classification is to benefit patients, families, and care givers who deal with ataxia. Having a name for one's ataxia provides a certain level of knowledge about the condition and guidelines for future testing and treatment.

To give you some perspective, the hereditary nature of ataxia has been known since the middle of the 19th century but it was not until late in the 20th century that the condition of ataxia was given a separate category in the 1967 edition of the International Classification of Diseases book, which is used to code diseases. There have been two editions since then which indicates the explosion in medical knowledge that has occurred during the past two decades. In the 1980's CT scan and MRI increased knowledge of ataxia and in the 1990's genetic research resulted in many breakthroughs for ataxia knowledge. In the early 2000's better ways were developed to diagnose multiple system atrophy (MSA) and sporadic ataxia.



**The exact classification of the sporadic degenerative ataxias may vary. These may be difficult to differentiate and may evolve over time.*

The inherited ataxias are named as follows:

Disease Type	Inheritance Pattern	Gene Test Available
SCA1 – Spinocerebellar Ataxia Type 1	Dominant	Yes
SCA2 – Spinocerebellar Ataxia Type 2	Dominant	Yes
SCA3/MJD – Spinocerebellar Ataxia Type 3/ Machado Joseph Disease	Dominant	Yes
SCA4 – Spinocerebellar Ataxia Type 4	Dominant	No
SCA5 – Spinocerebellar Ataxia Type 5	Dominant	Yes
SCA6 – Spinocerebellar Ataxia Type 6	Dominant	Yes
SCA7 – Spinocerebellar Ataxia Type 7	Dominant	Yes
SCA8 – Spinocerebellar Ataxia Type 8	Dominant	Yes
SCA10 – Spinocerebellar Ataxia Type 10	Dominant	Yes
SCA11 – Spinocerebellar Ataxia Type 11	Dominant	No
SCA12 – Spinocerebellar Ataxia Type 12	Dominant	Yes
SCA13 – Spinocerebellar Ataxia Type 13	Dominant	Yes
SCA14 – Spinocerebellar Ataxia Type 14	Dominant	Yes
SCA15 – Spinocerebellar Ataxia Type 15	Dominant	No
SCA16 – Spinocerebellar Ataxia Type 16	Dominant	No
SCA17 – Spinocerebellar Ataxia Type 17	Dominant	Yes
SCA18 – Spinocerebellar Ataxia Type 18	Dominant	No
SCA19 – Spinocerebellar Ataxia Type 19	Dominant	No
SCA20 – Spinocerebellar Ataxia Type 20	Dominant	No
SCA21 – Spinocerebellar Ataxia Type 21	Dominant	No
SCA22 – Spinocerebellar Ataxia Type 22	Dominant	No
SCA23 – Spinocerebellar Ataxia Type 23	Dominant	No
SCA25 – Spinocerebellar Ataxia Type 25	Dominant	No
SCA26 – Spinocerebellar Ataxia Type 26	Dominant	No
SCA27 – Spinocerebellar Ataxia Type 27	Dominant	No
SCA28 – Spinocerebellar Ataxia Type 28	Dominant	No
SCA29 – Spinocerebellar Ataxia Type 29	Dominant	No
DRPLA – Dentatorubral-Pallidoluyian Atrophy	Dominant	Yes
FXTAS – Fragile X Tremor-Ataxia	X-Linked	Yes
EA1 – Episodic Ataxia Type 1	Dominant	No
EA2 – Episodic Ataxia Type 2	Dominant	No
EA3 – Episodic Ataxia Type 3	Dominant	No
EA4 – Episodic Ataxia Type 4	Dominant	No
EA5 – Episodic Ataxia Type 5	Dominant	No
EA6 – Episodic Ataxia Type 6	Dominant	No
EA7 – Episodic Ataxia Type 7	Dominant	No
FRDA - Friedreich's Ataxia	Recessive	Yes
A-T -Ataxia Telangiectasia	Recessive	Yes
AOA1 and AOA2 – Ataxia with Oculomotor Apraxia Type 1 and Type 2	Recessive	Yes
ARSACS -Autosomal Recessive Ataxia of Charlevoix-Saguenay	Recessive	Yes
AVED -Ataxia, Friedreich like, with Vitamin Edeficiency (metabolic disorder)	Recessive	Yes
MIRAS-Mitochondrial Recessive Ataxia Syndrome	Recessive	Yes
MSS-Marinesco-SjögrenSyndrome	Recessive	Yes

A little history of the classification of ataxia may add some insight into this group of neurodegenerative diseases. Initially ataxia as a disease was categorized in two ways. If ataxia symptoms developed at an early age, it was diagnosed as Friedreich's ataxia. If ataxic symptoms developed after age 20, it was often given the name Marie's ataxia. Dominantly inherited ataxias were all called Marie's. Other forms of ataxia that were known by the early 20th century were sensory ataxias (ataxia caused by damage to nerves controlling sensation in the limbs) caused by vitamin B12 deficiency or syphilis infection.

Before the discovery of genes, five types of Olivopontocerebellar atrophy (OPCA) were classified based on autopsy, rather than age of onset or inheritance pattern. They were different from Late Onset Cerebellar Atrophy because other parts of the brain outside the cerebellum were involved. The letters O, P, and C refer to parts of the brain. O refers to the olive, P to the pons and C to the cerebellum. The letter A refers to atrophy which means to decrease in size. The five types were as follows:

- Menzel type (OPCA I)
- Fickler-Winkle or Dejerine-Thomas (OPCA II)
- OPCA with retinal degeneration (OPCA III)
- Schut-Haymaker (OPCA IV)
- OPCA with dementia and extrapyramidal signs (OPCA V)

Some of these OPCAs are now known to be genetic ataxias. Some have been renamed MSA, SND, or SDS, when Parkinson's disease-like symptoms or changes in blood pressure were present. Some are still called OPCA and research to find the cause is still ongoing.

Recently, many of the dominantly inherited ataxias have been renamed Spinocerebellar Ataxia or SCA. This is usually followed by a number which indicates the ataxia is caused by a specific genetic defect. The

numbers are usually assigned in the order in which the gene was identified. The SCA1 gene was found in 1993. The SCA29 gene was reported in 2008. As of 2008, genetic testing is available for twelve of the dominantly inherited ataxias: SCA1, SCA2, SCA3, SCA5, SCA6, SCA7, SCA8, SCA10, SCA12, SCA13, SCA14 and SCA17. Genetic testing is also available for DRPLA and FXTAS. Through research, one or two new genetic tests for dominant ataxia become available each year.

There are many other recessive ataxias that affect genetics or metabolism in nerve cells. Most are very rare and many do have genetic testing available. Please refer to the classification chart. You should work with your doctor in trying to identify the specific type of recessive ataxia that you or your child may have.

The National Ataxia Foundation (NAF) is committed to education about ataxia, service to individuals affected with the various forms of ataxia, and promoting and funding research to find the causes, better treatments, and a cure for ataxia. NAF can help by providing information for you, your family and your physician about ataxia. NAF does encourage you to visit our website at www.ataxia.org for additional information on ataxia, a listing of ataxia support groups, online chat groups, and more. For additional questions please contact NAF using the contact information listed below.

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