



## autosomal recessive cerebellar ataxia type 1

Autosomal recessive cerebellar ataxia type 1 (ARCA1) is a condition characterized by progressive problems with movement due to a loss (atrophy) of nerve cells in the part of the brain that coordinates movement (the cerebellum). Signs and symptoms of the disorder first appear in early to mid-adulthood. People with this condition initially experience impaired speech (dysarthria), problems with coordination and balance (ataxia), or both. They may also have difficulty with movements that involve judging distance or scale (dysmetria). Other features of ARCA1 include abnormal eye movements (nystagmus) and problems following the movements of objects with the eyes. The movement problems are slowly progressive, often resulting in the need for a cane, walker, or wheelchair.

### Frequency

More than 100 people have been diagnosed with ARCA1. This condition was first discovered in individuals from the Beauce and Bas-Saint-Laurent regions of Quebec, Canada, but it has since been found in populations worldwide.

### Genetic Changes

Mutations in the *SYNE1* gene cause ARCA1. The *SYNE1* gene provides instructions for making a protein called Syne-1 that is found in many tissues, but it seems to be especially critical in the brain. Within the brain, the Syne-1 protein appears to play a role in the maintenance of the cerebellum, which is the part of the brain that coordinates movement. The Syne-1 protein is active (expressed) in Purkinje cells, which are located in the cerebellum and are involved in chemical signaling between nerve cells (neurons).

*SYNE1* gene mutations that cause ARCA1 result in an abnormally short, dysfunctional version of the Syne-1 protein. The defective protein is thought to impair Purkinje cell function and disrupt signaling between neurons in the cerebellum. The loss of brain cells in the cerebellum causes the movement problems characteristic of ARCA1, but it is unclear how this cell loss is related to impaired Purkinje cell function.

### Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## Other Names for This Condition

- ARCA1
- autosomal recessive spinocerebellar ataxia 8
- recessive ataxia of Beauce

## Diagnosis & Management

These resources address the diagnosis or management of ARCA1:

- GeneReview: SYNE1-Related Autosomal Recessive Cerebellar Ataxia  
<https://www.ncbi.nlm.nih.gov/books/NBK1379>
- Genetic Testing Registry: Spinocerebellar ataxia, autosomal recessive 8  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853116/>
- Johns Hopkins Medicine Department of Neurology and Neurosurgery: What is Ataxia?  
[http://www.hopkinsmedicine.org/neurology\\_neurosurgery/centers\\_clinics/movement\\_disorders/ataxia/conditions/](http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/movement_disorders/ataxia/conditions/)
- MedlinePlus Encyclopedia: Dysarthria--Care  
<https://medlineplus.gov/ency/patientinstructions/000033.htm>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>

## Additional Information & Resources

### MedlinePlus

- Encyclopedia: Dysarthria--Care  
<https://medlineplus.gov/ency/patientinstructions/000033.htm>
- Encyclopedia: Movement--Uncoordinated  
<https://medlineplus.gov/ency/article/003198.htm>

- Health Topic: Balance Problems  
<https://medlineplus.gov/balanceproblems.html>
- Health Topic: Cerebellar Disorders  
<https://medlineplus.gov/cerebellardisorders.html>
- Health Topic: Movement Disorders  
<https://medlineplus.gov/movementdisorders.html>

#### Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Ataxias and Cerebellar or Spinocerebellar Degeneration Information Page  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Ataxias-and-Cerebellar-or-Spinocerebellar-Degeneration-Information-Page>

#### Educational Resources

- CLIMB: Cerebellar Ataxia Information Sheet  
<http://www.climb.org.uk/IMD/Charlie/CerebellarAtaxia.pdf>
- Disease InfoSearch: Spinocerebellar ataxia, autosomal recessive 8  
<http://www.diseaseinfosearch.org/Spinocerebellar+ataxia%2C+autosomal+recessive+8/9341>
- Johns Hopkins Medicine Department of Neurology and Neurosurgery: What is Ataxia?  
[http://www.hopkinsmedicine.org/neurology\\_neurosurgery/centers\\_clinics/movement\\_disorders/ataxia/conditions/](http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/movement_disorders/ataxia/conditions/)
- Kennedy Krieger Institute: Movement Disorders  
<https://www.kennedykrieger.org/patient-care/diagnoses-disorders/movement-disorders>
- Orphanet: Autosomal recessive ataxia, Beauce type  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=88644](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=88644)
- Washington University, St. Louis: Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/ataxia/recatax.html#ataxsyne1>

#### Patient Support and Advocacy Resources

- Ataxia UK  
<http://www.ataxia.org.uk/>
- CLIMB: Children Living with Inherited Metabolic Diseases  
<http://www.climb.org.uk/>

- National Ataxia Foundation  
<http://www.ataxia.org/>
- University of Kansas Medical Center Resource List: Ataxia  
<http://www.kumc.edu/gec/support/ataxia.html>

#### GeneReviews

- SYNE1-Related Autosomal Recessive Cerebellar Ataxia  
<https://www.ncbi.nlm.nih.gov/books/NBK1379>

#### Genetic Testing Registry

- Spinocerebellar ataxia, autosomal recessive 8  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853116/>

#### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22autosomal+recessive+cerebellar+ataxia+type+1%22+OR+%22Cerebellar+Ataxia%22>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SYNE1%5BTIAB%5D%29+AND+%28ataxia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

#### OMIM

- SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 8  
<http://omim.org/entry/610743>

### **Sources for This Summary**

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