SYNE1 gene

spectrin repeat containing nuclear envelope protein 1

Normal Function

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. The Syne-1 protein plays a role in the maintenance of the part of the brain that coordinates movement (the cerebellum). 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). The protein is thought to attach the membrane of Purkinje cells to the actin cytoskeleton, which is a network of fibers that make up the cell's structural framework. It is not clear what role this attachment plays in Purkinje cell function.

Health Conditions Related to Genetic Changes

Autosomal recessive cerebellar ataxia type 1

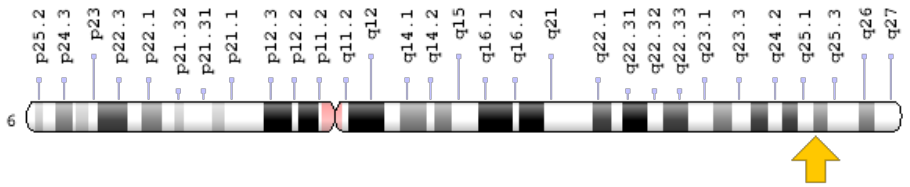
At least seven mutations in the *SYNE1* gene have been found to cause autosomal recessive cerebellar ataxia type 1 (ARCA1). All the mutations that have been identified create a premature stop signal in the instructions for making the Syne-1 protein, resulting in an abnormally short protein with impaired function. A dysfunctional Syne-1 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.

Emery-Dreifuss muscular dystrophy

Chromosomal Location

Cytogenetic Location: 6q25.2, which is the long (q) arm of chromosome 6 at position 25.2

Molecular Location: base pairs 152,121,684 to 152,637,395 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARCA1
- MYNE1
- myocyte nuclear envelope protein 1
- Nesp1
- nesprin-1
- nuclear envelope spectrin repeat protein 1
- spectrin repeat containing, nuclear envelope 1
- SYNE1_HUMAN

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Cerebellar Circuitry and the Coordination of Ongoing Movement
  https://www.ncbi.nlm.nih.gov/books/NBK10840/
- Washington University, St. Louis: Neuromuscular Disease Center
  https://neuromuscular.wustl.edu/ataxia/recatax.html#ataxsyne1

Clinical Information from GeneReviews

- SYNE1 Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1379

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SYNE1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SPECTRIN REPEAT-CONTAINING NUCLEAR ENVELOPE PROTEIN 1
  http://omim.org/entry/608441

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SYNE1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SYNE1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:17089
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:23345
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/23345
- UniProt
  https://www.uniprot.org/uniprot/Q8NF91

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  http://omim.org/entry/608441

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