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

MedlinePlus Genetics provides information about Emery-Dreifuss muscular dystrophy

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

Tests Listed in the Genetic Testing Registry

- Tests of SYNE1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23345[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; SYNE1 (https://omim.org/entry/608441)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/23345)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SYNE1[gene])

References

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- Dupre N, Gros-Louis F, Chrestian N, Verreault S, Brunet D, de Verteuil D,Brais B, Bouchard JP, Rouleau GA. Clinical and genetic study of autosomalrecessive cerebellar ataxia type 1. Ann Neurol. 2007 Jul;62(1):93-8. doi:10.1002/ana.21143. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17503513)
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1038/ng1927. Epub 2006 Dec 10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17159980)

Genomic Location

The *SYNE1* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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