SYNE1 Gene

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Spectrin repeat containing nuclear envelope protein 1: 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.

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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.

2. Health Conditions Related to Genetic Changes

2.1. 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.

2.2. Emery-Dreifuss muscular dystrophy

3. 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

References

1. Beaudin M, Gamache PL, Gros-Louis F, Dupré N. SYNE1 Deficiency. 2007 Feb 23[updated 2018 Dec 6]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH,Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): Universityof Washington, Seattle; 1993-2020. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1379/

2. Dupré N, Gros-Louis F, Chrestian N, Verreault S, Brunet D, de Verteuil D,Brais B, Bouchard JP, Rouleau GA. Clinical a nd genetic study of autosomalrecessive cerebellar ataxia type 1. Ann Neurol. 2007 Jul;62(1):93-8.
3. Gros-Louis F, Dupré N, Dion P, Fox MA, Laurent S, Verreault S, Sanes JR,Bouchard JP, Rouleau GA. Mutations in SY NE1 lead to a newly discovered form ofautosomal recessive cerebellar ataxia. Nat Genet. 2007 Jan;39(1):80-5.