BIN1 gene

bridging integrator 1

Normal Function

The *BIN1* gene provides instructions for making a protein that is found in tissues throughout the body, where it interacts with a variety of other proteins. The BIN1 protein is thought to be involved in the transportation of materials from the cell surface into the cell (endocytosis) and the self-destruction of cells (apoptosis). The BIN1 protein may act as a tumor suppressor protein, which means it prevents cells from growing and dividing too rapidly or in an uncontrolled way.

Several different versions (isoforms) of the BIN1 protein are produced from the *BIN1* gene. These isoforms vary by size and are active in different tissues. The BIN1 protein isoform that is expressed in muscle cells is thought to be involved in the formation of structures called transverse tubules or T tubules. These structures are found within the membrane of muscle cells, where they play a role in muscle tensing (contraction) and relaxation.

Health Conditions Related to Genetic Changes

Centronuclear myopathy

At least 10 mutations in the *BIN1* gene have been found to cause centronuclear myopathy, a condition that is characterized by muscle weakness (myopathy) in the skeletal muscles, which are the muscles used for movement. Most of these mutations change single protein building blocks (amino acids) in the BIN1 protein. *BIN1* gene mutations result in the production of a protein that cannot form T tubules. A shortage of T tubules in muscle fibers alters their structure, which prevents them from contracting and relaxing normally. The abnormal muscle fibers underlie the muscle weakness characteristic of centronuclear myopathy.

Other Names for This Gene

- AMPH2
- amphiphysin II
- amphiphysin-like protein
- AMPHL

- BIN1_HUMAN
- box-dependent myc-interacting protein 1
- myc box-dependent-interacting protein 1
- SH3P9

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of BIN1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=274[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28BIN1%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- BRIDGING INTEGRATOR 1; BIN1 (https://omim.org/entry/601248)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/274)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=BIN1[gene])

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Genomic Location

The *BIN1* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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