

LETM1 gene

leucine zipper and EF-hand containing transmembrane protein 1

Normal Function

The *LETM1* gene provides instructions for making a protein whose function is not well understood. This protein is active in mitochondria, which are structures within cells that convert the energy from food into a form that cells can use. The LETM1 protein may be involved in the transport of charged calcium atoms (calcium ions) across membranes within mitochondria. Researchers suspect that the protein also plays a role in determining the shape and volume of mitochondria.

Health Conditions Related to Genetic Changes

Wolf-Hirschhorn syndrome

The *LETM1* gene is located in a region of chromosome 4 that is deleted in people with the typical features of Wolf-Hirschhorn syndrome. The features of this condition include a characteristic facial appearance, delayed growth and development, intellectual disability, and seizures.

As a result of this deletion in chromosome 4, affected individuals are missing one copy of the *LETM1* gene in each cell. Studies suggest that a loss of this gene alters the structure of mitochondria; however, it is unclear how this abnormality is related to the signs and symptoms of Wolf-Hirschhorn syndrome. The deletion of the *LETM1* gene and other nearby genes can cause seizures or other abnormal electrical activity in the brain.

Other Names for This Gene

- LETM1_HUMAN
- leucine zipper-EF-hand containing transmembrane protein 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LETM1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3954\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3954[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LETM1%5BTIAB%5D%29+OR+%28leucine+zipper+AND+EF-hand+containing%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- LEUCINE ZIPPER/EF-HAND-CONTAINING TRANSMEMBRANE PROTEIN 1; LETM1 (<https://omim.org/entry/604407>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3954>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LETM1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LETM1[gene]))

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Genomic Location

The *LETM1* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

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