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. As a result of this deletion, 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. Specifically, a loss of the LETM1 gene has been associated with seizures or other abnormal electrical activity in the brain.

Chromosomal Location
Cytogenetic Location: 4p16.3, which is the short (p) arm of chromosome 4 at position 16.3
Molecular Location: base pairs 1,811,479 to 1,856,247 on chromosome 4 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

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

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/604407

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_LETM1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=LETM1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3954
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/O95202

Sources for This Summary

- Endele S, Fuhry M, Pak SJ, Zabel BU, Winterpacht A. LETM1, a novel gene encoding a putative EF-hand Ca(2+)-binding protein, flanks the Wolf-Hirschhorn syndrome (WHS) critical region and is deleted in most WHS patients. Genomics. 1999 Sep 1;60(2):218-25. Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10486213
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15138253

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14706454

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17696124

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18628306

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https://ghr.nlm.nih.gov/gene/LETM1

Reviewed: January 2009
Published: May 28, 2019

Lister Hill National Center for Biomedical Communications
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