EIF2B2 gene

eukaryotic translation initiation factor 2B subunit beta

Normal Function

The *EIF2B2* gene provides instructions for making one of five parts of a protein called eIF2B, specifically the beta subunit of this protein. The eIF2B protein helps regulate overall protein production (synthesis) in the cell by interacting with another protein, eIF2. The eIF2 protein is called an initiation factor because it is involved in starting (initiating) protein synthesis.

Under some conditions, eIF2B increases protein synthesis by helping to recycle molecules called GTP, which carry energy to the initiation factor. Under other conditions, it slows protein synthesis by binding tightly to the initiation factor, which converts the eIF2B protein into an inactive form and prevents recycling of GTP.

Proper regulation of protein synthesis is vital for ensuring that the correct levels of protein are available for the cell to cope with changing conditions. For example, cells must synthesize protein much faster if they are multiplying than if they are in a resting state.

Health Conditions Related to Genetic Changes

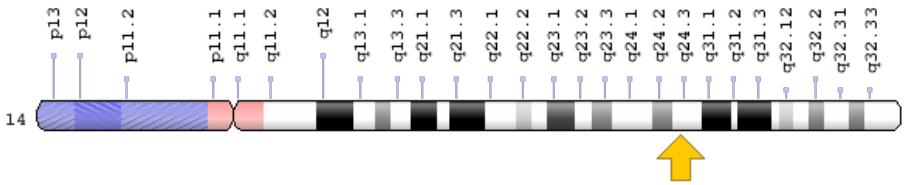
Leukoencephalopathy with vanishing white matter

Mutations in the *EIF2B2* gene have been identified in a few people with leukoencephalopathy with vanishing white matter, including some affected females with a variant of the disorder in which the neurological features are accompanied by ovarian failure (ovarioleukodystrophy). These mutations cause partial loss of eIF2B function. Impairment of eIF2B function makes it more difficult for the body's cells to regulate protein synthesis and deal with changing conditions and stress. Researchers believe that cells in the white matter (nerve fibers covered by a fatty substance called myelin that insulates and protects nerves) may be particularly affected by an abnormal response to stress, resulting in the signs and symptoms of leukoencephalopathy with vanishing white matter.

Chromosomal Location

Cytogenetic Location: 14q24.3, which is the long (q) arm of chromosome 14 at position 24.3

Molecular Location: base pairs 75,002,921 to 75,012,366 on chromosome 14 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EI2BB_HUMAN
- EIF-2Bbeta
- EIF2B
- eukaryotic translation initiation factor 2B, subunit 2 (beta, 39kD)
- eukaryotic translation initiation factor 2B, subunit 2 beta, 39kDa

Additional Information & Resources

Educational Resources

- Eurekah Bioscience: Mechanism of Translation Initiation in Eukaryotes https://www.ncbi.nlm.nih.gov/books/NBK6597/
- Molecular Biology of the Cell (fourth edition, 2002): The Phosphorylation of an Initiation Factor Globally Regulates Protein Synthesis https://www.ncbi.nlm.nih.gov/books/NBK26890/#A1387

Clinical Information from GeneReviews

- Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter https://www.ncbi.nlm.nih.gov/books/NBK1258

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28EIF2B2%5BTIAB%5D%29+OR+%28EIF2B%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+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 2
  http://omim.org/entry/606454

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EIF2B2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3258
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:8892
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8892
- UniProt
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https://ghr.nlm.nih.gov/gene/EIF2B2

Reviewed: October 2007
Published: September 10, 2019

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