EIF2B5 gene

eukaryotic translation initiation factor 2B subunit epsilon

Normal Function

The *EIF2B5* gene provides instructions for making one of five parts of a protein called eIF2B, specifically the epsilon 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 *EIF2B5* gene have been identified in about 65 percent of people with leukoencephalopathy with vanishing white matter, including those with a severe, early-onset form that is seen among the Cree and Chippewayan populations of Quebec and Manitoba (Cree leukoencephalopathy) and 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.

Other Names for This Gene

- CACH
- CLE
- EI2BE_HUMAN
- EIF-2B
- eIF-2B GDP-GTP exchange factor
- EIF2Bepsilon
- eukaryotic translation initiation factor 2B, subunit 5 (epsilon, 82kD)
- eukaryotic translation initiation factor 2B, subunit 5 epsilon, 82kDa
- LVWM

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EIF2B5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8893[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28EIF2B5%5BTIAB%5D%29+OR+%28%28CACH%5BTIAB%5D%29+OR+%28CLE%5BTIAB%5D%29+OR+%28EIF-2B%5BTIAB%5D%29+OR+%28EIF2Bepsilon%5BTIAB%5D%29+OR+%28LVWM%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 5; EIF2B5 (https://omim.org/entry/603945)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8893)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=EIF2B5[gene])

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Genomic Location

The *EIF2B5* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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