CLPB gene

ClpB family mitochondrial disaggregase

Normal Function

The *CLPB* gene provides instructions for making a protein whose function is unknown. The CLPB protein is found in cells throughout the body but is most abundant in the brain. Based on its similarity to a protein in other organisms, researchers speculate that the CLPB protein helps unfold misfolded proteins so they can be refolded correctly. When misfolded, proteins cannot function properly and may be damaging to cells.

Health Conditions Related to Genetic Changes

CLPB deficiency

At least 20 *CLPB* gene mutations have been found to cause CLPB deficiency. This condition is characterized by neurological problems, including movement abnormalities and seizures; a shortage of white blood cells (neutropenia), which can increase the risk of infections; and clouding of the lenses of the eyes (cataracts). In addition, affected individuals have an increased amount of a molecule called 3-methylglutaconic acid in their urine, which does not appear to cause health problems. The severity of these features varies widely. Many of the *CLPB* gene mutations lead to an abnormally short CLPB protein that is likely broken down quickly. Other mutations may reduce CLPB's function. The severity of the condition is thought to be related to the amount of functional protein remaining: severe CLPB deficiency is likely caused by a complete absence of CLPB protein, while moderate and mild CLPB deficiency result when some functional CLPB protein is produced. Researchers are unsure how reduction or absence of this protein leads to the signs and symptoms of CLPB deficiency.

Other Names for This Gene

- ANKCLB
- ankyrin-repeat containing bacterial clp fusion
- caseinolytic peptidase B protein homolog isoform 1
- caseinolytic peptidase B protein homolog isoform 2
- caseinolytic peptidase B protein homolog isoform 3
- caseinolytic peptidase B protein homolog isoform 4

- ClpB caseinolytic peptidase B homolog
- FLJ13152
- HSP78
- SKD3
- suppressor of potassium transport defect 3
- testicular secretory protein Li 11

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CLPB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=81570[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CLPB%5BTIAB%5D%29+OR+%28ClpB+homolog,+mitochondrial+AAA+ATPase+chaperonin%5BTIAB%5D%29%29+OR+%28%28ClpB+caseinolytic+peptidase+B+homolog%5BTIAB%5D%29+OR+%28HSP78%5BTIAB%5D%29+OR+%28SKD3%5BTIAB%5D%29+OR+%28ankyrin-repeat+containing+bacterial+clp+fusion%5BTIAB%5D%29+OR+%28caseinolytic+peptidase+B+protein+homolog+isoform+1%5BTIAB%5D%29+OR+%28caseinolytic+peptidase+B+protein+homolog+isoform+2%5BTIAB%5D%29+OR+%28caseinolytic+peptidase+B+protein+homolog+isoform+3%5BTIAB%5D%29+OR+%28caseinolytic+peptidase+B+protein+homolog+isoform+4%5BTIAB%5D%29+OR+%28suppressor+of+potassium+transport+defect+3%5BTIAB%5D%29+OR+%28testicular+secretory+protein+Li+11%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CASEINOLYTIC PEPTIDASE B; CLPB (https://omim.org/entry/616254)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/81570)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CLPB[gene])

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Genomic Location

The *CLPB* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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