UPB1 gene

beta-ureidopropionase 1

Normal Function

The *UPB1* gene provides instructions for making an enzyme called beta-ureidopropionase. This enzyme is involved in the breakdown of molecules called pyrimidines, which are building blocks of DNA and its chemical cousin RNA.

The beta-ureidopropionase enzyme is involved in the last step of the process that breaks down pyrimidines. This step converts N-carbamyl-beta-aminoisobutyric acid to beta-aminoisobutyric acid and also breaks down N-carbamyl-beta-alanine to beta-alanine, ammonia, and carbon dioxide. Both beta-aminoisobutyric acid and beta-alanine are thought to play roles in the nervous system. Beta-aminoisobutyric acid increases the production of a protein called leptin, which has been found to help protect brain cells from damage caused by toxins, inflammation, and other factors. Research suggests that beta-alanine is involved in sending signals between nerve cells (synaptic transmission) and in controlling the level of a chemical messenger (neurotransmitter) called dopamine.

Health Conditions Related to Genetic Changes

Beta-ureidopropionase deficiency

At least 16 *UPB1* gene mutations have been identified in people with beta-ureidopropionase deficiency. This disorder causes excessive amounts of N-carbamyl-beta-aminoisobutyric acid and N-carbamyl-beta-alanine to be released in the urine. Affected individuals may also have a variety of neurological problems such as seizures and intellectual disability, ranging from mild to severe. Some people with beta-ureidopropionase deficiency have no neurological symptoms, and the disorder can only be diagnosed with laboratory testing.

The mutations that cause beta-ureidopropionase deficiency reduce or eliminate beta-ureidopropionase enzyme activity. Loss of this enzyme function reduces the production of beta-aminoisobutyric acid and beta-alanine, and leads to an excess of their precursor molecules, N-carbamyl-beta-aminoisobutyric acid and N-carbamyl-beta-alanine, which are released in the urine. Reduced production of beta-aminoisobutyric acid and beta-alanine may impair their functions in the nervous system, leading to neurological problems in some people with beta-ureidopropionase deficiency. The extent of the

reduction in enzyme activity caused by a particular *UPB1* gene mutation, along with other genetic and environmental factors, may determine whether people with beta-ureidopropionase deficiency develop neurological problems and the severity of these problems.

Other Names for This Gene

- beta-alanine synthase
- beta-ureidopropionase
- BUP1
- n-carbamoyl-beta-alanine amidohydrolase
- ureidopropionase, beta

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of UPB1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51733[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28UPB1%5BTIAB%5D%29+OR+%28%28beta-ureidopropionase%5BTIAB%5D%29+OR+%28BUP1%5BTIAB%5D%29+OR+%28beta-alanine+synthase%5BTIAB%5D%29+OR+%28n-carbamoyl-beta-alanine+amidohydrolase%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)

Catalog of Genes and Diseases from OMIM

- BETA-UREIDOPROPIONASE; UPB1 (https://omim.org/entry/606673)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/51733)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=UPB1[gene])

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Genomic Location

The *UPB1* gene is found on chromosome 22 (https://medlineplus.gov/genetics/chromosome/22/).

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