CPS1 gene

carbamoyl-phosphate synthase 1

Normal Function

The *CPS1* gene provides instructions for making the enzyme carbamoyl phosphate synthetase I. This enzyme participates in the urea cycle, a series of reactions that occurs in liver cells. The urea cycle processes excess nitrogen, generated when protein is used by the body, into a compound called urea that is excreted by the kidneys. Excreting the excess nitrogen prevents it from accumulating in the form of ammonia, which is toxic.

The specific role of carbamoyl phosphate synthetase I is to control the first step of the urea cycle, a reaction in which excess nitrogen compounds are incorporated into the cycle to be processed.

Health Conditions Related to Genetic Changes

Carbamoyl phosphate synthetase I deficiency

Approximately 10 mutations that cause carbamoyl phosphate synthetase I deficiency have been identified in the *CPS1* gene. A mutated *CPS1* gene may result in a carbamoyl phosphate synthetase I enzyme that is shorter than normal or the wrong shape, or may prevent the enzyme from being produced at all.

The shape of an enzyme affects its ability to control a chemical reaction. If the carbamoyl phosphate synthetase enzyme is misshapen or missing, it cannot fulfill its role in the urea cycle. Excess nitrogen is not converted to urea for excretion, and ammonia accumulates in the body. Ammonia is toxic, especially to the nervous system, so this accumulation causes neurological problems and other signs and symptoms of carbamoyl phosphate synthetase I deficiency.

Other disorders

One common alteration (polymorphism) in the *CPS1* gene has been associated with increased risk of circulatory problems in newborns and in individuals who have received bone marrow transplants. This genetic change results in the amino acid (protein building block) asparagine being substituted for the amino acid threonine at position 1405 in the protein sequence (written as Thr1405Asn or T1405N).

Researchers believe that this polymorphism in the *CPS1* gene may reduce the production of a compound called nitric oxide (NO). Normally, nitric oxide causes blood vessels to expand (dilate), which reduces blood pressure. A reduced amount of nitric oxide could lead to circulatory problems.

Other Names for This Gene

- carbamoyl phosphate synthase I
- carbamoyl-phosphate synthase 1, mitochondrial
- carbamoyl-phosphate synthase, mitochondrial precursor
- carbamoylphosphate synthetase I
- CPSase I
- CPSM_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CPS1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1373[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CPS1%5BTIAB%5D%29+OR+%28carbamoyl-phosphate+synthetase+1,+mitochondrial%5BTIAB%5D%29%29+OR+%28carbamoylphosphate+synthetase+I%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CARBAMOYL PHOSPHATE SYNTHETASE I; CPS1 (https://omim.org/entry/608307)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1373)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CPS1[gene])

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Genomic Location

The *CPS1* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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