

CBS gene

cystathionine beta-synthase

Normal Function

The *CBS* gene provides instructions for making an enzyme called cystathionine beta-synthase. This enzyme acts in a chemical pathway and is responsible for using vitamin B6 to convert building block of proteins (amino acid) called homocysteine and serine to a molecule called cystathionine. Another enzyme then converts cystathionine to the amino acid cysteine, which is used to build proteins or is broken down and excreted in urine. Additionally, other amino acids, including methionine, are produced in this pathway.

Health Conditions Related to Genetic Changes

Homocystinuria

More than 150 mutations that cause homocystinuria have been identified in the *CBS* gene. Most of these mutations change single amino acids in cystathionine beta-synthase. The most common mutation substitutes the amino acid threonine for the amino acid isoleucine at position 278 in the enzyme (written as Ile278Thr or I278T). Another common mutation, which is the most frequent cause of homocystinuria in the Irish population, replaces the amino acid glycine with the amino acid serine at position 307 (written as Gly307Ser or G307S). These mutations disrupt the normal function of cystathionine beta-synthase. As a result, homocysteine and other potentially toxic compounds build up in the blood, and homocysteine is excreted in urine. Researchers have not determined how excess homocysteine leads to the signs and symptoms of homocystinuria.

Other Names for This Gene

- beta-thionase
- CBS_HUMAN
- HIP4
- methylcysteine synthase
- serine sulfhydrase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CBS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=875\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=875[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CBS%5BTIAB%5D%29+OR+%28cystathionine-beta-synthase%5BTIAB%5D%29%29+AND+%28%28I-serine+hydro-lyase+%28adding+homocysteine%29%29+OR+%28beta-thionase%5BMAJR%5D%29+OR+%28serine+sulfhydrase%5BMAJR%5D%29+OR+%28cystathionine+synthetase%5BMAJR%5D%29+OR+%28cystathionine+beta-synthase%5BMAJR%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

- CYSTATHIONINE BETA-SYNTHASE; CBS (<https://omim.org/entry/613381>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/875>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CBS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CBS[gene]))

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Genomic Location

The *CBS* gene is found on chromosome 21 (<https://medlineplus.gov/genetics/chromosome/21/>).

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