

BCHE gene

butyrylcholinesterase

Normal Function

The *BCHE* gene provides instructions for making the pseudocholinesterase enzyme, also known as butyrylcholinesterase, which is produced by the liver and circulates in the blood. The pseudocholinesterase enzyme is involved in the breakdown of certain drugs, including muscle relaxant drugs called choline esters that are used during general anesthesia. These drugs are given to relax the muscles used for movement (skeletal muscles), including the muscles involved in breathing, and are often employed in emergencies when a breathing tube must be inserted quickly.

Pseudocholinesterase also helps protect the body by breaking down certain toxic substances before they reach the nerves. These substances include certain pesticides, poisons that attack the nerves, and specific natural toxins including a compound called solanine found in green potato skin. It is likely that the enzyme has other functions in the body, but these functions are not well understood. Studies suggest that the enzyme may be involved in the transmission of nerve signals.

Health Conditions Related to Genetic Changes

Pseudocholinesterase deficiency

More than 50 mutations in the *BCHE* gene have been identified in people with pseudocholinesterase deficiency, a condition that results in increased sensitivity to choline esters and certain other drugs. Some of these mutations replace single protein building blocks (amino acids) in the pseudocholinesterase enzyme, resulting in an abnormal enzyme that does not function properly. Other mutations prevent the production of pseudocholinesterase. A lack (deficiency) of functional pseudocholinesterase enzyme impairs the body's ability to break down choline ester drugs efficiently, leading to abnormally prolonged drug effects.

Other Names for This Gene

- acylcholine acylhydrolase
- butyrylcholine esterase
- CHE1

- CHLE_HUMAN
- choline esterase II
- cholinesterase
- cholinesterase 1
- cholinesterase precursor
- E1
- pseudocholinesterase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of BCHE ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=590\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=590[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28BCHE%5BTIAB%5D%29+OR+%28butyrylcholinesterase%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+1080+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- BUTYRYLCHOLINESTERASE; BCHE (<https://omim.org/entry/177400>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/590>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=BCHE\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=BCHE[gene]))

References

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Genomic Location

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

Last updated April 1, 2012