# DBH gene

dopamine beta-hydroxylase

## Normal Function

The *DBH* gene provides instructions for producing the enzyme dopamine beta (β)-hydroxylase. This enzyme converts dopamine to norepinephrine, both of which are chemical messengers (neurotransmitters) that transmit signals between nerve cells. Norepinephrine plays an important role in the autonomic nervous system, which controls involuntary body processes such as the regulation of blood pressure and body temperature.

## Health Conditions Related to Genetic Changes

### Dopamine beta-hydroxylase deficiency

At least six mutations in the *DBH* gene have been found to cause dopamine β-hydroxylase deficiency. The most common mutation (usually written as IVS1+2T>C) interferes with the normal processing of dopamine β-hydroxylase. As a result of this mutation, an abnormally short, nonfunctional version of the enzyme is produced. A lack of functional dopamine β-hydroxylase leads to a shortage of norepinephrine, which causes difficulty with regulating blood pressure and other autonomic nervous system problems seen in dopamine β-hydroxylase deficiency.

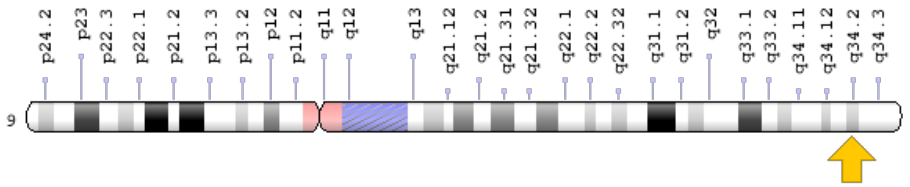
### Other disorders

Studies have shown certain variations (polymorphisms) in the *DBH* gene to be associated with increased risk of attention deficit hyperactivity disorder (ADHD). *DBH* gene polymorphisms are also thought to increase the risk of psychotic symptoms in people with schizophrenia or unipolar major depression. Other studies, however, have not supported these findings. Many genetic and environmental factors are believed to contribute to these complex conditions.

## Chromosomal Location

Cytogenetic Location: 9q34.2, which is the long (q) arm of chromosome 9 at position 34.2

Molecular Location: base pairs 133,636,363 to 133,659,344 on chromosome 9 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- DBM
- dopamine beta-hydroxylase (dopamine beta-monooxygenase)
- dopamine beta-monooxygenase
- DOPO_HUMAN

## Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Biosynthesis of Catecholamines https://www.ncbi.nlm.nih.gov/books/NBK27988/
- Endocrinology (first edition, 2001): Biosynthesis and control of catecholamines secreted by the adrenal medulla https://www.ncbi.nlm.nih.gov/books/NBK26/?rendertype=box&id=A467
- Endocrinology (first edition, 2001): Catecholamine synthesis and secretion https://www.ncbi.nlm.nih.gov/books/NBK26/#A698
- Neuroscience (second edition, 2001): The Biogenic Amines https://www.ncbi.nlm.nih.gov/books/NBK11035/

Clinical Information from GeneReviews

- Dopamine Beta-Hydroxylase Deficiency https://www.ncbi.nlm.nih.gov/books/NBK1474

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DBH%5BTIAB%5D%29+OR+%28dopamine+beta-hydroxylase%5BTI%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

Catalog of Genes and Diseases from OMIM

- DOPAMINE BETA-HYDROXYLASE, PLASMA
  http://omim.org/entry/609312

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DBH.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DBH%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2689
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1621
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1621
- UniProt
  https://www.uniprot.org/uniprot/P09172

## Sources for This Summary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  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/15088079
- OMIM: DOPAMINE BETA-HYDROXYLASE, PLASMA
  http://omim.org/entry/609312
- Kim CH, Zabetian CP, Cubells JF, Cho S, Biaggioni I, Cohen BM, Robertson D, Kim KS. Mutations in the dopamine beta-hydroxylase gene are associated with human norepinephrine deficiency. Am J Med Genet. 2002 Mar 1;108(2):140-7.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/11857564
- Vincent S, Robertson D. The broader view: catecholamine abnormalities. Clin Auton Res. 2002 May;12 Suppl 1:I44-9.
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