DBH gene
dopamine beta-hydroxylase

Normal Function

The $DBH$ gene provides instructions for producing the enzyme dopamine beta ($\beta$)-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 $\beta$-hydroxylase deficiency. The most common mutation (usually written as IVS1+2T>C) interferes with the normal processing of dopamine $\beta$-hydroxylase. As a result of this mutation, an abnormally short, nonfunctional version of the enzyme is produced. A lack of functional dopamine $\beta$-hydroxylase leads to a shortage of norepinephrine, which causes difficulty with regulating blood pressure and other autonomic nervous system problems seen in dopamine $\beta$-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 133,659,329 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20191205, GRCh38.p13) (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

• Endocrinology (first edition, 2001): Catecholamine synthesis and secretion

• 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
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1621
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P09172

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15088079
- OMIM: DOPAMINE BETA-HYDROXYLASE, PLASMA
  http://omim.org/entry/609312
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11857564
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12102462