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.

Other Names for This Gene

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

- DOPO_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DBH (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1621[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; DBH (https://omim.org/entry/609312)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1621)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DBH[gene])

References

- Cubells JF, Zabetian CP. Human genetics of plasma dopamine beta-hydroxylaseactivity: applications to research in psychiatry and neurology. Psychopharmacology (Berl). 2004 Aug;174(4):463-76. doi:10.1007/s00213-004-1840-8. Epub 2004 Apr 16. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15088079)
- 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 humannorepinephrine deficiency. Am J Med Genet. 2002 Mar 1;108(2):140-7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11857564)
- Vincent S, Robertson D. The broader view: catecholamine abnormalities. ClinAuton Res. 2002 May;12 Suppl 1:I44-9. doi: 10.1007/s102860200018. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12102462)

Genomic Location

The *DBH* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

Last updated September 1, 2008