



## DDC gene

dopa decarboxylase

### Normal Function

The *DDC* gene provides instructions for making the aromatic l-amino acid decarboxylase (AADC) enzyme, which is important in the brain and nervous system. This enzyme takes part in the pathway that produces dopamine and serotonin, which are chemical messengers that transmit signals between nerve cells (neurotransmitters).

Dopamine is produced from the protein building block (amino acid) tyrosine, and serotonin is produced from the amino acid tryptophan. Both neurotransmitters are produced in two-step processes. First, other enzymes control the reactions that convert tyrosine to L-dopa, and tryptophan to 5-hydroxytryptophan. The AADC enzyme then converts L-dopa and 5-hydroxytryptophan to dopamine and serotonin, respectively. To do this, it removes a molecular structure called a carboxyl group, consisting of a carbon atom, two oxygen atoms, and a hydrogen atom.

### Health Conditions Related to Genetic Changes

#### Aromatic l-amino acid decarboxylase deficiency

Mutations in the *DDC* gene result in reduced activity of the AADC enzyme. Without enough of this enzyme, nerve cells produce less dopamine and serotonin. Dopamine and serotonin are necessary for normal nervous system function, and changes in the levels of these neurotransmitters contribute to the developmental delay, intellectual disability, abnormal movements, and autonomic dysfunction seen in people with AADC deficiency.

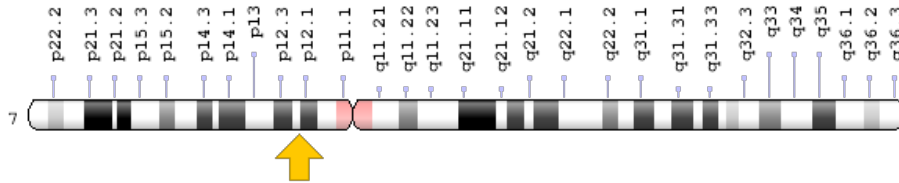
#### Other disorders

Studies have shown certain variations (polymorphisms) in the *DDC* gene to be associated with increased risk of nicotine dependence, schizophrenia, bipolar disorder, and attention deficit hyperactivity disorder; however, other studies have not supported these findings. Many genetic and environmental factors are believed to contribute to these complex conditions.

## Chromosomal Location

Cytogenetic Location: 7p12.2-p12.1, which is the short (p) arm of chromosome 7 between positions 12.2 and 12.1

Molecular Location: base pairs 50,458,436 to 50,565,460 on chromosome 7 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AADC
- aromatic L-amino acid decarboxylase
- dopa decarboxylase (aromatic L-amino acid decarboxylase)

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Dopa Decarboxylase Catalyzes the Removal of the Carboxyl Group from DOPA to Form Dopamine  
<https://www.ncbi.nlm.nih.gov/books/NBK27988/#A862>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DDC%5BTIAB%5D%29+OR+%28dopa+decarboxylase%5BTIAB%5D%29%29+OR+%28%28AADC%5BTIAB%5D%29+OR+%28aromatic+L-amino+acid+decarboxylase%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>

### Catalog of Genes and Diseases from OMIM

- DOPA DECARBOXYLASE  
<http://omim.org/entry/107930>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/DDCID50590ch7p12.html>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=DDC%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:2719](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:2719)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:1644>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1644>
- UniProt  
<https://www.uniprot.org/uniprot/P20711>

## **Sources for This Summary**

- OMIM: DOPA DECARBOXYLASE  
<http://omim.org/entry/107930>
- Hyland K, Surtees RA, Rodeck C, Clayton PT. Aromatic L-amino acid decarboxylase deficiency: clinical features, diagnosis, and treatment of a new inborn error of neurotransmitter amine synthesis. *Neurology*. 1992 Oct;42(10):1980-8.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/1357595>
- Hyland K. Inherited disorders affecting dopamine and serotonin: critical neurotransmitters derived from aromatic amino acids. *J Nutr*. 2007 Jun;137(6 Suppl 1):1568S-1572S; discussion 1573S-1575S.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17513427>
- Pearl PL, Capp PK, Novotny EJ, Gibson KM. Inherited disorders of neurotransmitters in children and adults. *Clin Biochem*. 2005 Dec;38(12):1051-8. Epub 2005 Nov 18. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16298354>
- Pearl PL, Taylor JL, Trzcinski S, Sokohl A. The pediatric neurotransmitter disorders. *J Child Neurol*. 2007 May;22(5):606-16. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17690069>
- Verbeek MM, Geurtz PB, Willemsen MA, Wevers RA. Aromatic L-amino acid decarboxylase enzyme activity in deficient patients and heterozygotes. *Mol Genet Metab*. 2007 Apr;90(4):363-9. Epub 2007 Jan 19.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17240182>
- Yu Y, Panhuysen C, Kranzler HR, Hesselbrock V, Rounsaville B, Weiss R, Brady K, Farrer LA, Gelernter J. Intronic variants in the dopa decarboxylase (DDC) gene are associated with smoking behavior in European-Americans and African-Americans. *Hum Mol Genet*. 2006 Jul 15;15(14):2192-9. Epub 2006 Jun 1.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16740595>
- Zhang H, Ye Y, Wang X, Gelernter J, Ma JZ, Li MD. DOPA decarboxylase gene is associated with nicotine dependence. *Pharmacogenomics*. 2006 Dec;7(8):1159-66.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17184203>

---

Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/DDC>

Reviewed: May 2008

Published: May 14, 2019

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services