

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 (ADHD); however, other studies have not supported these findings. Many genetic and environmental factors are believed to contribute to these complex conditions.

Other Names for This Gene

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DDC ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1644\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1644[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- DOPA DECARBOXYLASE; DDC (<https://omim.org/entry/107930>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1644>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DDC\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DDC[gene]))

References

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Genomic Location

The *DDC* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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