DRD5 gene

dopamine receptor D5

Normal Function

The *DRD5* gene provides instructions for making a protein called dopamine receptor D5, which is found in the brain. This protein works together with a chemical messenger (neurotransmitter) called dopamine. Dopamine fits into the D5 receptor like a key in a lock, which triggers chemical reactions within nerve cells. Dopamine performs many important roles in the brain, including regulating attention, mood, memory, learning, and movement.

Health Conditions Related to Genetic Changes

Benign essential blepharospasm

Several studies have examined a possible relationship between a common variation (polymorphism) in the *DRD5* gene and benign essential blepharospasm. The results of these studies have been mixed. Some research has suggested that the polymorphism, a short segment of DNA known as allele 2 that repeats multiple times within the gene, occurs more often in people with benign essential blepharospasm than in people without the disorder. However, other studies have found no connection between this polymorphism and benign essential blepharospasm. Researchers are still working to clarify whether variants in the *DRD5* gene are associated with this disorder.

Other disorders

Other polymorphisms in the *DRD5* gene appear to be associated with a common behavioral condition called attention-deficit/hyperactivity disorder (ADHD). This condition, which typically begins in childhood, is characterized by overactivity, impulsive behavior, and difficulty paying attention.

Most studies of the *DRD5* gene and ADHD have focused on a polymorphism located near the beginning of the gene. The region consists of two DNA building blocks (base pairs) that are repeated multiple times in a row. The size of this segment ranges from 134 to 156 base pairs. Multiple studies have suggested that a particular polymorphism, which is 148-base pairs long, is associated with a moderately increased risk of ADHD. However, it is unclear how this polymorphism affects the risk of the disorder.

Polymorphisms in the *DRD5* gene are among the many factors that are being studied to help explain the causes of ADHD. A large number of genetic and environmental factors, most of which remain unknown, likely determine a person's risk of developing this complex condition.

Other Names for This Gene

- D1beta dopamine receptor
- DBDR
- dopamine D5 receptor
- dopamine receptor D1B
- DRD1B
- DRD1L2
- DRD5_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DRD5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1816[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DRD5%5BTIAB%5D%29+OR+%28dopamine+receptor+D5%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- DOPAMINE RECEPTOR D5; DRD5 (https://omim.org/entry/126453)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1816)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DRD5[gene])

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Genomic Location

The *DRD5* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

Last updated November 21, 2024