

ADCY5 gene

adenylate cyclase 5

Normal Function

The *ADCY5* gene provides instructions for making an enzyme called adenylate cyclase 5. This enzyme helps convert a molecule called adenosine triphosphate (ATP) to another molecule called cyclic adenosine monophosphate (cAMP). ATP is a molecule that supplies energy for cells' activities, including muscle contraction, and cAMP is involved in signaling for many cellular functions.

Health Conditions Related to Genetic Changes

ADCY5-related dyskinesia

At least six *ADCY5* gene mutations have been identified in people with *ADCY5*-related dyskinesia, a disorder that causes abnormal involuntary movements. These mutations are thought to enhance adenylate cyclase 5 enzyme activity and lead to higher levels of cAMP within cells, so they are described as "gain-of-function" mutations. Other *ADCY5* gene mutations prevent production of adenylate cyclase 5. It is unclear how either type of mutation leads to the abnormal movements that occur in this disorder.

Other disorders

Certain normal variations (polymorphisms) in the *ADCY5* gene have been associated with an increased risk of type 2 diabetes, which is the most common form of diabetes and results in impaired control of blood glucose, also called blood sugar. cAMP normally increases in response to increases in blood glucose and is involved in signaling that stimulates the production of insulin. Researchers suggest that the polymorphisms associated with increased type 2 diabetes risk may decrease the ability of the adenylate cyclase 5 enzyme to produce cAMP, resulting in the abnormal response to glucose that occurs in type 2 diabetes.

Other Names for This Gene

- AC5
- adenylate cyclase type 5
- adenylate cyclase type 5 isoform 1

- adenylate cyclase type 5 isoform 2
- adenylate cyclase type V
- adenylyl cyclase 5
- ATP pyrophosphate-lyase 5
- FDFM

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ADCY5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=111\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=111[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ADCY5%5BTIAB%5D%29+OR+%28adenylate+cyclase+5%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%29%29%29>)

Catalog of Genes and Diseases from OMIM

- ADENYLATE CYCLASE 5; ADCY5 (<https://omim.org/entry/600293>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/111>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ADCY5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ADCY5[gene]))

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Genomic Location

The *ADCY5* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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