

## ADCY5-related dyskinesia

### Description

*ADCY5*-related dyskinesia is a movement disorder; the term "dyskinesia" refers to abnormal involuntary movements. The abnormal movements that occur in *ADCY5*-related dyskinesia typically appear as sudden (paroxysmal) jerks, twitches, tremors, muscle tensing (dystonia), or writhing (choreiform) movements, and can affect the limbs, neck, and face.

The abnormal movements associated with *ADCY5*-related dyskinesia usually begin between infancy and late adolescence. They can occur continually during waking hours, and frequently also disturb sleep. The involuntary movements often occur when changing position, such as from sitting to standing, or when deliberately making other movements.

Severely affected infants may experience weak muscle tone (hypotonia) and delay in development of motor skills such as crawling and walking; later, these individuals may have difficulties with activities of daily living and may eventually require a wheelchair. In more mildly affected individuals, the condition has little impact on walking and other motor skills, although the abnormal movements can lead to clumsiness or difficulty with social acceptance in school or other situations.

In some people with *ADCY5*-related dyskinesia, the disorder is generally stable throughout their lifetime. In others, it slowly gets worse (progresses) in both frequency and severity before stabilizing or even improving in middle age. Anxiety, fatigue, and other stress can temporarily increase the severity of the signs and symptoms of *ADCY5*-related dyskinesia, while some affected individuals may experience remission periods of days or weeks without abnormal movements. Life expectancy is not usually affected by *ADCY5*-related dyskinesia, and most people with this condition have normal intelligence.

### Frequency

At least 400 people have been diagnosed with *ADCY5*-related dyskinesia, but its prevalence is unknown. The disorder is thought to be underdiagnosed because its features can resemble those of other conditions such as cerebral palsy or epilepsy.

## Causes

As its name suggests, *ADCY5*-related dyskinesia is caused by mutations in the *ADCY5* gene. This 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. Some *ADCY5* gene mutations that cause *ADCY5*-related dyskinesia are thought to increase adenylate cyclase 5 enzyme activity and the level of cAMP within cells. Others prevent production of adenylate cyclase 5. It is unclear how either type of mutation leads to the abnormal movements that occur in this disorder.

[Learn more about the gene associated with ADCY5-related dyskinesia](#)

- *ADCY5*

## Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

## Other Names for This Condition

- Familial dyskinesia with facial myokymia
- FDFM

## Additional Information & Resources

### Genetic Testing Information

- Genetic Testing Registry: Dyskinesia with orofacial involvement, autosomal dominant (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1847627/>)

### Genetic and Rare Diseases Information Center

- Familial dyskinesia and facial myokymia (<https://rarediseases.info.nih.gov/diseases/12722/index>)

### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

### Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22ADCY5-related dyskinesia%22](https://clinicaltrials.gov/search?cond=%22ADCY5-related+dyskinesia%22))

### Catalog of Genes and Diseases from OMIM

- DYSKINESIA WITH OROFACIAL INVOLVEMENT, AUTOSOMAL DOMINANT; DSKOD (<https://omim.org/entry/606703>)

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28dyskinesia%5BTIAB%5D%29+AND+%28%28myokymia%5BALL%5D%29+OR+%28adcy5%5BALL%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

## **References**

- Carapito R, Paul N, Untrau M, Le Gentil M, Ott L, Alsaleh G, Jochem P, Radosavljevic M, Le Caignec C, David A, Damier P, Isidor B, Bahram S. A de novo ADCY5 mutation causes early-onset autosomal dominant chorea and dystonia. *MovDisord.* 2015 Mar;30(3):423-7. doi: 10.1002/mds.26115. Epub 2014 Dec 27. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25545163>)
- Chen DH, Meneret A, Friedman JR, Korvatska O, Gad A, Bonkowski ES, Stessman HA, Doummar D, Mignot C, Anheim M, Bernes S, Davis MY, Damon-Perriere N, Degos B, Grabli D, Gras D, Hisama FM, Mackenzie KM, Swanson PD, Tranchant C, Vidailhet M, Winesett S, Trouillard O, Amendola LM, Dorschner MO, Weiss M, Eichler EE, Torkamani A, Roze E, Bird TD, Raskind WH. ADCY5-related dyskinesia: Broaderspectrum and genotype-phenotype correlations. *Neurology.* 2015 Dec8;85(23):2026-35. doi: 10.1212/WNL.0000000000002058. Epub 2015 Nov 4. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/26537056>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4676753/>)
- Chen YZ, Friedman JR, Chen DH, Chan GC, Bloss CS, Hisama FM, Topol SE, Carson AR, Pham PH, Bonkowski ES, Scott ER, Lee JK, Zhang G, Oliveira G, Xu J, Scott-VanZeeland AA, Chen Q, Levy S, Topol EJ, Storm D, Swanson PD, Bird TD, Schork NJ, Raskind WH, Torkamani A. Gain-of-function ADCY5 mutations in familial dyskinesia with facial myokymia. *Ann Neurol.* 2014 Apr;75(4):542-9. doi: 10.1002/ana.24119. Epub 2014 Mar 13. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24700542>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4457323/>)
- Chen YZ, Matsushita MM, Robertson P, Rieder M, Girirajan S, Antonacci F, Lipe H,

Eichler EE, Nickerson DA, Bird TD, Raskind WH. Autosomal dominant familial dyskinesia and facial myokymia: single exome sequencing identifies a mutation in adenylyl cyclase 5. *Arch Neurol*. 2012 May;69(5):630-5. doi:10.1001/archneurol.2012.54. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22782511>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3508680/>)

- Mencacci NE, Erro R, Wiethoff S, Hersheson J, Ryten M, Balint B, Ganos C, Stamelou M, Quinn N, Houlden H, Wood NW, Bhatia KP. ADCY5 mutations are another cause of benign hereditary chorea. *Neurology*. 2015 Jul 7;85(1):80-8. doi:10.1212/WNL.0000000000001720. Epub 2015 Jun 17. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/26085604>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4501937/>)
- Raskind WH, Matsushita M, Peter B, Biberston J, Wolff J, Lipe H, Burbank R, Bird TD. Familial dyskinesia and facial myokymia (FDFM): Follow-up of a large family and linkage to chromosome 3p21-3q21. *Am J Med Genet B Neuropsychiatr Genet*. 2009 Jun 5;150B(4):570-4. doi: 10.1002/ajmg.b.30879. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18980218>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3116722/>)

**Last updated August 1, 2017**