ADCY5-related dyskinesia

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 in some cases also during sleep. Severely affected infants may experience weak muscle tone (hypotonia) and delay in development of motor skills such as crawling and walking; 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 and intelligence are unaffected by this disorder.

Frequency

The prevalence of *ADCY5*-related dyskinesia is unknown. At least 50 affected individuals have been described in the medical literature.

Genetic Changes

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.

Inheritance Pattern

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

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Dyskinesia, familial, with facial myokymia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1847627/

Other Diagnosis and Management Resources

- GeneReview: ADCY5-Related Dyskinesia
  https://www.ncbi.nlm.nih.gov/books/NBK263441

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Movement -- Uncontrollable
  https://medlineplus.gov/ency/article/003201.htm
- Health Topic: Movement Disorders
  https://medlineplus.gov/movementdisorders.html

Genetic and Rare Diseases Information Center

- ADCY5-related dyskinesia
  https://rarediseases.info.nih.gov/diseases/12722/adcy5-related-dyskinesia

Educational Resources

- MalaCards: adcy5-related dyskinesia
  http://www.malacards.org/card/adcy5_related_dyskinesia
- Merck Manual Consumer Version: Overview of Movement Disorders
  http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/movement-disorders/overview-of-movement-disorders
- Orphanet: Familial dyskinesia and facial myokymia
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=324588

Patient Support and Advocacy Resources

- ADCY5.org
  http://www.adcy5.org/home.html
- Dystonia Medical Research Foundation: Paroxysmal Dyskinesias
  https://www.dystonia-foundation.org/what-is-dystonia/forms-of-dystonia/paroxysmal-dyskinesias
- National Ataxia Foundation
  http://ataxia.org/

GeneReviews

- ADCY5-Related Dyskinesia
  https://www.ncbi.nlm.nih.gov/books/NBK263441

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

OMIM

- DYSKINESIA, FAMILIAL, WITH FACIAL MYOKYMIA
  http://omim.org/entry/606703

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