

Myoclonus-dystonia

Description

Myoclonus-dystonia is a movement disorder that typically affects the neck, torso, and arms. Individuals with this condition experience quick, involuntary muscle jerks or twitches (myoclonus). About half of individuals with myoclonus-dystonia develop dystonia, which is involuntary tensing of various muscles that causes unusual positioning. In myoclonus-dystonia, dystonia often affects one or both hands, causing writer's cramp, or the neck, causing the head to turn (torticollis).

The movement problems usually first appear in childhood or early adolescence with the development of myoclonus. In most cases, the movement problems remain stable throughout life. In some adults, myoclonus improves with alcohol consumption, which can lead to affected individuals self-medicating and developing alcohol use disorder.

People with myoclonus-dystonia often develop psychological disorders such as depression, anxiety, panic attacks, and obsessive-compulsive disorder (OCD).

Frequency

The prevalence of myoclonus-dystonia in Europe is estimated to be 1 in 500,000 individuals. Its prevalence elsewhere in the world is unknown.

Causes

Mutations in the *SGCE* gene cause 30 to 50 percent of cases of myoclonus-dystonia. The *SGCE* gene provides instructions for making a protein called epsilon (ϵ)-sarcoglycan, whose function is unknown. The ϵ -sarcoglycan protein is located within the outer membrane of cells in many tissues, but it is most abundant in nerve cells (neurons) in the brain and in muscle cells.

SGCE gene mutations that cause myoclonus-dystonia result in a shortage (deficiency) of functional ϵ -sarcoglycan protein. This lack of functional protein seems to affect the regions of the brain involved in coordinating and controlling movements (the cerebellum and basal ganglia, respectively). It is unknown why *SGCE* gene mutations seem to affect only these areas of the brain.

Mutations in multiple other genes are associated with myoclonus-dystonia. Mutations in each of these genes cause a small percentage of cases. These genes are primarily

active (expressed) in the brain and mutations likely lead to impairment of normal movement.

Some people with myoclonus-dystonia do not have an identified mutation in any of the known associated genes. The cause of the condition in these individuals is unknown.

[Learn more about the genes associated with Myoclonus-dystonia](#)

- RELN
- SGCE

Additional Information from NCBI Gene:

- KCTD17

Inheritance

In cases in which the genetic cause is known, myoclonus-dystonia 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 cases in which the cause of the condition is unknown, the inheritance is unclear.

When caused by *SGCE* gene mutations, myoclonus-dystonia occurs only when the mutation is inherited from a person's father. People normally inherit one copy of each gene from their mother and one copy from their father. For most genes, both copies are active, or "turned on," in all cells. For a small subset of genes, however, only one of the two copies is active. For some of these genes, only the copy inherited from a person's father (the paternal copy) is active, while for other genes, only the copy inherited from a person's mother (the maternal copy) is active. These differences in gene activation based on the gene's parent of origin are caused by a phenomenon called genomic imprinting.

Because only the paternal copy of the *SGCE* gene is active, myoclonus-dystonia occurs when mutations affect the paternal copy of the *SGCE* gene. Mutations in the maternal copy of the gene typically do not cause any health problems. Rarely, individuals who inherit an *SGCE* gene mutation from their mothers will develop features of myoclonus-dystonia. It is unclear why a gene that is supposed to be turned off is active in these rare cases.

Other genes associated with myoclonus-dystonia are not imprinted, and mutations that cause the condition can be inherited from either parent.

Other Names for This Condition

- Dystonia 11
- DYT11
- Myoclonus-dystonia syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Myoclonic dystonia 11 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1834570/>)

Genetic and Rare Diseases Information Center

- Myoclonus-dystonia syndrome (<https://rarediseases.info.nih.gov/diseases/7139/index>)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22Myoclonus-dystonia%22>)

Catalog of Genes and Diseases from OMIM

- DYSTONIA 11, MYOCLONIC; DYT11 (<https://omim.org/entry/159900>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28myoclonus-dystonia%5BTIAB%5D%29+OR+%28myoclonic+dystonia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

References

- Esapa CT, Waite A, Locke M, Benson MA, Kraus M, McIlhinney RA, Sillitoe RV, Beesley PW, Blake DJ. SGCE missense mutations that cause myoclonus-dystonia syndrome impair epsilon-sarcoglycan trafficking to the plasma membrane: modulation by ubiquitination and torsinA. *Hum Mol Genet.* 2007 Feb 1;16(3):327-42. doi: 10.1093/hmg/ddl472. Epub 2007 Jan 2. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17200151>)
- Gerrits MC, Foncke EM, Koelman JH, Tijssen MA. Pediatric writer's cramp in myoclonus-dystonia: maternal imprinting hides positive family history. *Eur J Paediatr Neurol.* 2009 Mar;13(2):178-80. doi: 10.1016/j.ejpn.2008.03.007. Epub 2008 Jun 20. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18571946>)

- Groen JL, Ritz K, Jalalzadeh H, van der Salm SM, Jongejan A, Mook OR, Haagsmans MA, Zwinderman AH, Motazacker MM, Hennekam RC, Baas F, Tijssen MA. RELN rare variants in myoclonus-dystonia. *Mov Disord*. 2015 Mar;30(3):415-9. doi:10.1002/mds.26070. Epub 2015 Feb 4. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25648840>)
- Mencacci NE, Rubio-Agusti I, Zdebik A, Asmus F, Ludtmann MH, Ryten M, Plagnol V, Hauser AK, Bandres-Ciga S, Bettencourt C, Forabosco P, Hughes D, Soutar MM, Peall K, Morris HR, Trabzuni D, Tekman M, Stanescu HC, Kleta R, Carecchio M, Zorzi G, Nardocci N, Garavaglia B, Lohmann E, Weissbach A, Klein C, Hardy J, Pittman AM, Foltynie T, Abramov AY, Gasser T, Bhatia KP, Wood NW. A missense mutation in KCTD17 causes autosomal dominant myoclonus-dystonia. *Am J Hum Genet*. 2015 Jun 4;96(6):938-47. doi: 10.1016/j.ajhg.2015.04.008. Epub 2015 May 14. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25983243>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4457957/>)
- Nardocci N, Zorzi G, Barzaghi C, Zibordi F, Ciano C, Ghezzi D, Garavaglia B. Myoclonus-dystonia syndrome: clinical presentation, disease course, and genetic features in 11 families. *Mov Disord*. 2008 Jan;23(1):28-34. doi:10.1002/mds.21715. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17853490>)
- Peall KJ, Kurian MA, Wardle M, Waite AJ, Hedderly T, Lin JP, Smith M, Whone A, Pall H, White C, Lux A, Jardine PE, Lynch B, Kirov G, O'Riordan S, Samuel M, Lynch T, King MD, Chinnery PF, Warner TT, Blake DJ, Owen MJ, Morris HR. SGCE and myoclonus dystonia: motor characteristics, diagnostic criteria and clinical predictors of genotype. *J Neurol*. 2014 Dec;261(12):2296-304. doi:10.1007/s00415-014-7488-3. Epub 2014 Sep 11. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25209853>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4495322/>)
- Ritz K, Gerrits MC, Foncke EM, van Ruissen F, van der Linden C, Vergouwen MD, Bloem BR, Vandenberghe W, Crols R, Speelman JD, Baas F, Tijssen MA. Myoclonus-dystonia: clinical and genetic evaluation of a large cohort. *J Neurol Neurosurg Psychiatry*. 2009 Jun;80(6):653-8. doi: 10.1136/jnnp.2008.162099. Epub 2008 Dec 9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19066193>)

Last updated October 1, 2017