Myoclonus-dystonia

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.

Inheritance Pattern

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

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Myoclonic dystonia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1834570/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22myoclonus-dystonia%22

Other Diagnosis and Management Resources

- GeneReview: SGCE Myoclonus-Dystonia
  https://www.ncbi.nlm.nih.gov/books/NBK1414

Additional Information & Resources

Health Information from MedlinePlus

- Health Topic: Dystonia
  https://medlineplus.gov/dystonia.html
- Health Topic: Movement Disorders
  https://medlineplus.gov/movementdisorders.html

Genetic and Rare Diseases Information Center

- Myoclonus-dystonia
  https://rarediseases.info.nih.gov/diseases/7139/myoclonus-dystonia

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Dystonias Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Dystonias-Information-Page
- National Institute of Neurological Disorders and Stroke: Myoclonus Fact Sheet
  https://www.ninds.nih.gov/Disorders/All-Disorders/Myoclonus-Information-Page

Educational Resources

- Kennedy Krieger Institute: Movement Disorders
  https://www.kennedykrieger.org/patient-care/conditions/movement-disorders
- Merck Manual Consumer Version: Dystonia
  https://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/movement-disorders/dystonia
- Merck Manual Consumer Version: Myoclonus
  https://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/movement-disorders/myoclonus
- Orphanet: Myoclonus-dystonia syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=36899

Patient Support and Advocacy Resources

- Dystonia Medical Research Foundation
  https://dystonia-foundation.org/
- International Parkinson and Movement Disorder Society: Myoclonus and Startle
  https://www.movementdisorders.org/MDS/About/Movement-Disorder-Overviews/Myoclonus--Startle.htm

- The Dystonia Society (UK)
  https://www.dystonia.org.uk/
- The Global Dystonia Registry
  https://www.globaldystoniaregistry.org/

Clinical Information from GeneReviews

- SGCE Myoclonus-Dystonia
  https://www.ncbi.nlm.nih.gov/books/NBK1414

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- DYSTONIA 11, MYOCLONIC
  http://omim.org/entry/159900

Medical Genetics Database from MedGen

- Myoclonic dystonia
  https://www.ncbi.nlm.nih.gov/medgen/331778

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/myoclonus-dystonia

Reviewed: October 2017
Published: September 10, 2019

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services