Early-onset isolated dystonia

Description

Early-onset isolated dystonia is one of many forms of dystonia, which is a group of conditions characterized by involuntary tensing of the muscles (muscle contractions), twisting of specific body parts such as an arm or a leg, rhythmic shaking (tremors), and other uncontrolled movements. An isolated dystonia is one that occurs without other abnormal movements or other neurological symptoms, such as seizures, a loss of intellectual function, or developmental or intellectual delay. Early-onset isolated dystonia does not affect a person's intelligence.

The signs and symptoms of early-onset isolated dystonia tend to occur in mid-childhood or adolescence. Abnormal muscle spasms in an arm or a leg are usually the first sign. These unusual movements initially occur while a person is doing a specific action, such as writing or walking. In some affected people, dystonia later spreads to other parts of the body and the movements may become persistent and present when at rest and not doing an activity. The abnormal movements persist throughout life, but they do not usually cause pain.

The signs and symptoms of early-onset isolated dystonia vary from person to person, even among affected members of the same family. The mildest cases affect only a single part of the body, causing isolated problems such as abnormal posture and spasms of the hand while attempting to write (writer's cramp). Severe cases involve abnormal movements affecting many parts of the body.

Frequency

Early-onset isolated dystonia is among the most common forms of childhood dystonia. This disorder occurs most frequently in people of Ashkenazi (central and eastern European) Jewish heritage, affecting 1 in 3,000 to 9,000 people in this population. The condition is less common among people with other backgrounds. It is estimated to affect 1 in 10,000 to 30,000 non-Jewish people worldwide.

Causes

A particular variant (also called a mutation) in the *TOR1A* gene (also known as *DYT1*) is responsible for most cases of early-onset isolated dystonia. Variants in other genes cause other forms of dystonia, such as dystonia 6.

The *TOR1A* gene provides instructions for making a protein called torsinA. Although little is known about its function, this protein may help process and transport other proteins within cells. It appears to be critical for the normal development and function of nerve cells in the brain.

A variant in the *TOR1A* gene alters the structure of torsinA. The altered protein's effect on the function of nerve cells in the brain is unclear. People with early-onset isolated dystonia do not have a loss of nerve cells or obvious changes in the structure of the brain that would explain the abnormal muscle contractions. Instead, the altered torsinA protein may have subtle effects on the connections between nerve cells and likely disrupts chemical signaling between nerve cells that control movement. Researchers are working to determine how a change in this protein leads to the characteristic features of this disorder.

Learn more about the gene associated with Early-onset isolated dystonia

- TOR1A

Inheritance

Variants in the *TOR1A* gene are inherited in an autosomal dominant pattern, which means one of the two copies of the gene is altered in each cell. Many people who have a variant in this gene are not affected by the disorder and may never know they have the altered gene. Only 30 to 40 percent of people who inherit a *TOR1A* gene variant will ever develop signs and symptoms of early-onset isolated dystonia.

The vast majority of those who have been diagnosed with early-onset isolated dystonia have inherited a *TOR1A* variant from one parent. The parent may or may not have signs and symptoms of the condition, and other family members may or may not be affected.

In very rare cases, early-onset isolated dystonia is inherited in an autosomal recessive pattern, which means that both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Dystonia musculorum deformans 1
- DYT1
- Early-onset generalized torsion dystonia
- Early-onset primary dystonia
- Oppenheim dystonia
- Oppenheim's dystonia
- Primary torsion dystonia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Early-onset generalized limb-onset dystonia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1851945/)

Genetic and Rare Diseases Information Center

- Early-onset generalized limb-onset dystonia (https://rarediseases.info.nih.gov/diseases/2027/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- DYSTONIA 1, TORSION, AUTOSOMAL DOMINANT; DYT1 (https://omim.org/entry/128100)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=(Dystonic+Disorders%5BMAJR%5D)+AND+(early-onset+isolated+dystonia%5BTIAB%5D)+OR+(early-onset+primary+dystonia%5BTIAB%5D)+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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Last updated April 22, 2022