TOR1A gene

torsin family 1 member A

Normal Function

The *TOR1A* gene (also known as *DYT1*) provides instructions for making a protein called torsinA. This protein is found in the space between two neighboring structures within cells, the nuclear envelope and the endoplasmic reticulum. The nuclear envelope surrounds the nucleus and separates it from the rest of the cell. The endoplasmic reticulum processes proteins and other molecules and helps transport them to specific destinations either inside or outside the cell. Although little is known about the function of torsinA, studies suggest that it may help process and transport other proteins. TorsinA may also participate in the movement of membranes associated with the nuclear envelope and endoplasmic reticulum, and in stress response signaling.

TorsinA is active in many of the body's tissues, and it is particularly important for the normal function of nerve cells in the brain. For example, researchers have found high levels of torsinA in a part of the brain called the substantia nigra. This region contains nerve cells that produce dopamine, a chemical messenger that transmits signals within the brain to produce smooth physical movements.

Health Conditions Related to Genetic Changes

Early-onset isolated dystonia

A particular variant (also called a mutation) in the *TOR1A* gene causes most cases of early-onset isolated dystonia. This condition 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.

This *TOR1A* gene variant, which is often called the GAG deletion or delta GAG, deletes three DNA building blocks (base pairs) from the *TOR1A* gene. The resulting torsinA protein is missing one protein building block (amino acid) in a critical region. 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 seen with this condition. 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 and sensory feedback in the brain. Researchers are working to determine how a change in this protein leads to the characteristic features of early-onset isolated dystonia.

Benign essential blepharospasm

Several studies have examined a possible relationship between common variations (polymorphisms) in the *TOR1A* gene and several forms of adult-onset dystonia, most commonly dystonia isolated to a certain body region (task-specific focal dystonia), abnormal posture and spasms of the hand while attempting to write (writer's cramp), and spasms of the eyelids in the form of benign essential blepharospasm. The results of these studies have been mixed. Some research has suggested that certain polymorphisms increase a person's risk of developing these forms of dystonia. However, other studies have found no connection between changes in the *TOR1A* gene and condition risk. Researchers are still working to clarify whether variants of the *TOR1A* gene are related to adult-onset dystonias.

Other Names for This Gene

- DQ2
- Dystonia 1 protein
- dystonia 1, torsion (autosomal dominant; torsinA)
- DYT1
- TOR1A_HUMAN
- torsin family 1, member A (torsin A)
- Torsin-1A
- torsinA

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TOR1A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1861[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TOR1A%5BTIAB%5D%29+OR+%28%28DYT1%5BTIAB%5D%29+OR+%28torsin+A%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TORSIN 1A; TOR1A (https://omim.org/entry/605204)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1861)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TOR1A[gene])

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Genomic Location

The *TOR1A* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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