

TOR1A Gene

Subjects: **Genetics & Heredity**

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Torsin family 1 member A: The TOR1A gene (also known as *DYT1*) provides instructions for making a protein called torsinA.

genes

1. 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.

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.

2. Health Conditions Related to Genetic Changes

2.1. Early-onset primary dystonia

A particular mutation in the *TOR1A* gene causes most cases of early-onset primary dystonia. This mutation, 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 primary 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. Researchers are working to determine how a change in this protein leads to the characteristic features of this disorder.

2.2. 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, including 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 benign essential blepharospasm. However, other studies have found no connection between changes in the *TOR1A* gene and risk of this disorder. Researchers are still working to clarify whether variants of the *TOR1A* gene are related to benign essential blepharospasm and other adult-onset dystonias.

3. 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

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