# Benign Essential Blepharospasm

Subjects: Genetics

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## Definition

Benign essential blepharospasm is a condition characterized by abnormal blinking or spasms of the eyelids. This condition is a type of dystonia, which is a group of movement disorders involving uncontrolled tensing of the muscles (muscle contractions), rhythmic shaking (tremors), and other involuntary movements. Benign essential blepharospasm is different from the common, temporary eyelid twitching that can be caused by fatigue, stress, or caffeine.

## 1. Introduction

The signs and symptoms of benign essential blepharospasm usually appear in mid- to late adulthood and gradually worsen. The first symptoms of the condition include an increased frequency of blinking, dry eyes, and eye irritation that is aggravated by wind, air pollution, sunlight, and other irritants. These symptoms may begin in one eye, but they ultimately affect both eyes. As the condition progresses, spasms of the muscles surrounding the eyes cause involuntary winking or squinting. Affected individuals have increasing difficulty keeping their eyes open, which can lead to severe vision impairment.

In more than half of all people with benign essential blepharospasm, the symptoms of dystonia spread beyond the eyes to affect other facial muscles and muscles in other areas of the body. When people with benign essential blepharospasm also experience involuntary muscle spasms affecting the tongue and jaw (oromandibular dystonia), the combination of signs and symptoms is known as Meige syndrome.

## 2. Frequency

Benign essential blepharospasm affects an estimated 20,000 to 50,000 people in the United States. For unknown reasons, it occurs in women more than twice as often as it occurs in men.

## 3. Causes

The causes of benign essential blepharospasm are unknown, although the disorder likely results from a combination of genetic and environmental factors. Certain genetic changes probably increase the likelihood of developing this condition, and environmental factors may trigger the signs and symptoms in people who are at risk.

Studies suggest that this condition may be related to other forms of adult-onset dystonia, including uncontrolled twisting of the neck muscles (spasmodic torticollis) and spasms of the hand and finger muscles (writer's cramp). Researchers suspect that benign essential blepharospasm and similar forms of dystonia are associated with malfunction of the basal ganglia, which are structures deep within the brain that help start and control movement.

Although genetic factors are almost certainly involved in benign essential blepharospasm, no genes have been clearly associated with the condition. Several studies have looked at the relationship between common variations (polymorphisms) in the *DRD5* and *TOR1A* genes and the risk of developing benign essential blepharospasm. These studies have had conflicting results, with some showing an association and others finding no connection. Researchers are working to determine which genetic factors are related to this disorder.

### 3.1. The Genes Associated with Benign Essential Blepharospasm

- DRD5
- TOR1A

## 4. Inheritance

Most cases of benign essential blepharospasm are sporadic, which means that the condition occurs in people with no

history of this disorder or other forms of dystonia in their family.

Less commonly, benign essential blepharospasm has been found to run in families. In some of these families, the condition appears to have an autosomal dominant pattern of inheritance, which means that one copy of an altered gene in each cell is sufficient to cause the disorder. However, no causative genes have been identified.

## 5. Other Names for This Condition

- essential blepharospasm
- eyelid twitching
- primary blepharospasm
- spasm of eyelids

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### Keywords


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