Isolated Duane Retraction Syndrome

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Isolated Duane retraction syndrome is a disorder of eye movement.

Keywords: genetic conditions

1. Introduction

This condition prevents outward movement of the eye (toward the ear), and in some cases may also limit inward eye movement (toward the nose). As the eye moves inward, the eyelids partially close and the eyeball pulls back (retracts) into its socket. Most commonly, only one eye is affected. About 10 percent of people with isolated Duane retraction syndrome develop amblyopia ("lazy eye"), a condition that causes vision loss in the affected eye.

About 70 percent of all cases of Duane retraction syndrome are isolated, which means they occur without other signs and symptoms. Duane retraction syndrome can also occur as part of syndromes that affect other areas of the body. For example, Duane-radial ray syndrome is characterized by this eye disorder in conjunction with abnormalities of bones in the arms and hands.

Researchers have identified three forms of isolated Duane retraction syndrome, designated types I, II, and III. The types vary in which eye movements are most severely restricted (inward, outward, or both). All three types are characterized by retraction of the eyeball as the eye moves inward.

2. Frequency

Isolated Duane retraction syndrome affects an estimated 1 in 1,000 people worldwide. This condition accounts for 1 percent to 5 percent of all cases of abnormal eye alignment (strabismus). For unknown reasons, isolated Duane syndrome affects females more often than males.

3. Causes

In most people with isolated Duane retraction syndrome, the cause of the condition is unknown. However, researchers have identified mutations in one gene, *CHN1*, that cause the disorder in a small number of families. The *CHN1* gene provides instructions for making a protein that is involved in the early development of the nervous system. Specifically, the protein appears to be critical for the formation of nerves that control several of the muscles surrounding the eyes (extraocular muscles). Mutations in the *CHN1* gene disrupt the normal development of these nerves and the extraocular muscles needed for side-to-side eye movement. Abnormal function of these muscles leads to restricted eye movement and related problems with vision.

3.1. The gene associated with Isolated Duane retraction syndrome

- CHN1

4. Inheritance

Isolated Duane retraction syndrome usually occurs in people with no history of the disorder in their family. These cases are described as simplex, and their genetic cause is unknown.

Less commonly, isolated Duane retraction syndrome can run in families. Familial cases most often have an autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell is sufficient to cause the disorder. When isolated Duane retraction syndrome is caused by *CHN1* mutations, it has an autosomal dominant inheritance pattern.

In a few families with isolated Duane retraction syndrome, the pattern of affected family members suggests autosomal recessive inheritance. In these families, one or more children are affected, although the parents typically have no signs or symptoms of the condition. The parents of children with an autosomal recessive condition are called carriers, which means they carry one mutated copy of a gene in each cell. In affected children, both copies of the gene in each cell are mutated. However, researchers have not discovered the gene or genes responsible for autosomal recessive isolated Duane retraction syndrome.

5. Other Names for This Condition

- co-contractive retraction syndrome
- Duane anomaly, isolated
- Duane retraction syndrome
- Duane syndrome
- Duane's syndrome
- ocular retraction syndrome
- Stilling-Turk-Duane syndrome

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