



Duane syndrome

Other Names: Duane retraction syndrome; Stilling-Turk-Duane syndrome; Isolated Duane anomaly

Duane syndrome is a rare condition characterized by abnormal alignment of the eyes, and restriction of horizontal movement of the eyes. It is sometimes caused by a mutation in the CHN1 gene that affects growth of the nerves connected to the eye muscles.

Characteristics of Duane syndrome

Duane syndrome is a congenital (from birth) eye condition that affects an individual's ability to move and align their eyes. It typically results in strabismus (abnormal alignment of the eyes) and can cause amblyopia (lazy eye). It accounts for about 1-5% of total cases of strabismus and affects approximately 1 in 1,000 people throughout the world.

There are three main subtypes of Duane syndrome. Type 1 is characterized by an inability to move the eye outward (toward the ear) and accounts for about 75% of cases. Type 2 is characterized by an inability to move the eye inward (toward the nose) and accounts for about 10% of cases. Type 3 is characterized by problems with both moving the eye inward and outward and accounts for about 15% percent of cases. All three types can be unilateral (involve one eye) or bilateral (involve both eyes). Duane syndrome also causes retraction and/or unwanted vertical movements of the affected eye(s) when attempting to look toward the nose. Due to the misalignment of the eyes, individuals with Duane syndrome often develop a head tilt to correct their vision.

Approximately 70% of cases of Duane syndrome are isolated and have no other associated birth defects. Some individuals do have other congenital abnormalities, specifically of the ears, kidney, heart, upper limbs, and skeleton. One such condition that involves Duane syndrome and other anomalies is Duane-radial-ray syndrome. In addition to eye problems, this syndrome can also result in hand and finger abnormalities, deafness, and kidney problems.

Diagnosis/Testing

Duane syndrome is typically diagnosed based on clinical findings. Changes or mutations in the CHN1 gene have been identified in families with multiple individuals with isolated Duane syndrome (no other abnormalities). This gene plays a role in nerve cell development. The chance of detecting a mutation in the CHN1 gene is higher for individuals with bilateral Duane syndrome and for individuals with a family history of Duane syndrome.

Management/Surveillance

Management of Duane syndrome typically involves regular ophthalmology visits to monitor symptoms, treat amblyopia, and prevent further complications. Some individuals may need special glasses or contact lenses. In extreme cases, eye surgery may be warranted to help improve head tilt.

Individuals with additional findings should also undergo regular evaluation to monitor their other symptoms as indicated.

Mode of inheritance

For many individuals with Duane syndrome, they are the only affected person in their family. In these cases, the genetic cause is usually not identified and the mode of inheritance cannot be accurately predicted.

For individuals with an affected parent who has a mutation in the CHN1 gene, the condition is inherited in an autosomal dominant pattern. This means inheriting one CHN1 mutation is enough for an individual to be affected and show signs of Duane syndrome. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with Duane syndrome has a parent affected with the condition. Parents of an affected child should undergo ophthalmologic evaluation to look for a milder presentation of the condition. If a parent also has Duane syndrome, the risk of having a child with Duane syndrome is 50% with each pregnancy. If a parent does not have Duane syndrome, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Boston Children's Hospital: Duane Syndrome

<http://www.childrenshospital.org/health-topics/conditions/duane-syndrome>

Genetics Home Reference: Isolated Duane Retraction Syndrome

<http://ghr.nlm.nih.gov/condition/isolated-duane-retraction-syndrome>

Genetics Home Reference: Duane-radial ray syndrome

<http://ghr.nlm.nih.gov/condition/duane-radial-ray-syndrome>

References

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