Isolated Duane retraction syndrome

Description

Isolated Duane retraction syndrome is a disorder of eye movement. This condition prevents outward movement of the eye (toward the ear), and in some cases it may also limit inward eye movement (toward the nose). In people with this condition, the eyeball pulls back (retracts) into its socket and the eyelid opening narrows as the eye moves inward. Due to these movement limitations, the eyes often do not look in the same direction (strabismus). Instead, affected individuals may need to turn their head to track objects with both eyes.

Normally, only one eye is affected (most commonly the left eye) in people with isolated Duane retraction syndrome. About 10 percent of people with this condition also 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. The remaining cases of Duane retraction syndrome occur as part of syndromes that affect other areas of the body. For example, Duane-radial ray syndrome is characterized by this eye disorder and 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 are differentiated by which eye movements are most severely restricted (inward, outward, or both). All three types are characterized by a retraction of the eyeball as the eye moves inward.

Frequency

Isolated Duane retraction syndrome is estimated to affect 1 in 1,000 to 10,000 people worldwide. This condition accounts for 1 percent to 5 percent of all cases of strabismus.

Causes

In most people with isolated Duane retraction syndrome, the cause of the condition is unknown. However, researchers have identified variants (also called mutations) in the *CHN1* gene that cause the disorder in some affected individuals.

The *CHN1* gene provides instructions for making a protein called N-chimaerin. This protein is involved in the early development of the nervous system. Specifically, the

protein appears to be critical for the formation of nerves that send movement signals to several of the muscles surrounding the eyes (extraocular muscles). Variants in the *CHN1* gene disrupt the normal development of these nerves so they cannot effectively control the extraocular muscles. Impaired function of these muscles leads to restricted eye movement and related problems with vision.

Variants in other genes can cause isolated Duane retraction syndrome in a small number of people.

Learn more about the gene associated with Isolated Duane retraction syndrome

- CHN1

Additional Information from NCBI Gene:

- MAFB

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* gene variants, it has an autosomal dominant inheritance pattern.

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

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Duane retraction syndrome (https://www.ncbi.nlm.nih.gov/

gtr/conditions/C0013261/)

Genetic and Rare Diseases Information Center

- Duane retraction syndrome (https://rarediseases.info.nih.gov/diseases/6288/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Isolated Duane retracti on syndrome%22)

Catalog of Genes and Diseases from OMIM

- DUANE RETRACTION SYNDROME 1; DURS1 (https://omim.org/entry/126800)
- DUANE RETRACTION SYNDROME 2; DURS2 (https://omim.org/entry/604356)
- DUANE RETRACTION SYNDROME 3 WITH OR WITHOUT DEAFNESS; DURS3 (https://omim.org/entry/617041)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28Duane*+retraction+syndr ome%5BTIAB%5D%29+OR+%28Duane*+syndrome%5BTIAB%5D%29+OR+%28D URS2+%5BTIAB%5D+AND+Duane+%5BTIAB%5D%29%29+AND+english%5Bla% 5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

References

- Appukuttan B, Gillanders E, Juo SH, Freas-Lutz D, Ott S, Sood R, Van Auken A, Bailey-Wilson J, Wang X, Patel RJ, Robbins CM, Chung M, Annett G, Weinberg K, Borchert MS, Trent JM, Brownstein MJ, Stout JT. Localization of a gene for Duaneretraction syndrome to chromosome 2q31. Am J Hum Genet. 1999 Dec;65(6): 1639-46.doi: 10.1086/302656. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/ 10577917) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/arti cles/PMC1288374/)
- Barry BJ, Whitman MC, Hunter DG, Engle EC. Duane Syndrome. 2007 May 25[updated 2019 Aug 29]. In: Adam MP, Bick S, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, editors. GeneReviews(R) [Internet]. Seattle (WA): Universityof Washington, Seattle; 1993-2025. Available fromhttp://www.ncbi.nlm.nih.gov/books/

NBK1190/ Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/20301369)

- Demer JL, Clark RA, Lim KH, Engle EC. Magnetic resonance imaging evidence forwidespread orbital dysinnervation in dominant Duane's retraction syndrome linkedto the DURS2 locus. Invest Ophthalmol Vis Sci. 2007 Jan;48(1):194-202. doi: 10.1167/iovs.06-0632. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17197533) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1850629/)
- Engle EC, Andrews C, Law K, Demer JL. Two pedigrees segregating Duane'sretraction syndrome as a dominant trait map to the DURS2 genetic locus. InvestOphthalmol Vis Sci. 2007 Jan;48(1):189-93. doi: 10.1167/iovs.06-0631. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17197532) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2829295/)
- Miyake N, Chilton J, Psatha M, Cheng L, Andrews C, Chan WM, Law K, Crosier M, Lindsay S, Cheung M, Allen J, Gutowski NJ, Ellard S, Young E, Iannaccone A, Appukuttan B, Stout JT, Christiansen S, Ciccarelli ML, Baldi A, Campioni M,Zenteno JC, Davenport D, Mariani LE, Sahin M, Guthrie S, Engle EC. Human CHN1mutations hyperactivate alpha2-chimaerin and cause Duane's retraction syndrome.Science. 2008 Aug 8;321(5890):839-43. doi: 10.1126/science.1156121. Epub 2008 Jul24. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18653847) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2593867/)
- Mohan K, Sharma A, Pandav SS. Differences in epidemiological and clinicalcharacteristics between various types of Duane retraction syndrome in 331patients. J AAPOS. 2008 Dec;12(6):576-80. doi: 10.1016/j.jaapos.2008.04.015. Epub2008 Aug 23. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18723379)
- Prasad P, Saxena A, Saxena R. Duane syndrome: An overview on the currentmanagement. Taiwan J Ophthalmol. 2023 Dec 20;13(4):489-499. doi:10.4103/tjo.TJO-D-23-00078. eCollection 2023 Oct-Dec. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/38249504)

Last updated May 31, 2024