



Duane Retraction Syndrome 1

Alternative Names

DURS1
DRS
Duane Syndrome
DUS
Duane Anomaly
Retraction Syndrome

Record Category

Disease phenotype

WHO-ICD

N.B.: Classification not applicable to gene loci.

Incidence per 100,000 Live Births

51-100

OMIM Number

126800

Mode of Inheritance

Autosomal dominant

Gene Map Locus

8q13

Description

Duane retraction syndrome is a congenital disorder of eye movement, characterized by an inability of the affected eye to move outwards (abduction). In some cases, this might also be manifested in an ability of the eye to move inwards towards the nose (adduction). The condition is also associated with a retraction of the eyeball into the socket with attempted adduction, and a widening of the palpebral fissures on attempted abduction. There are three subtypes of Duane retraction syndrome; DRS Type 1 is characterized by restricted inwards movement, while Type 2 is characterized by restricted outward movement. Type 3 patients have both movements affected. In a large majority of the cases, the condition is unilateral affecting only one of the eyes. This is usually the left eye, in most cases. The underlying etiology of DRS is a defect in some of the eye muscles (specifically the lateral

and medial rectus muscles) receiving proper signal from the cranial nerves that innervate them.

DRS is a fairly common disorder, affecting 1 in 1,000 people worldwide, and accounts for 1-5% of all cases of strabismus. This condition is usually found isolated. However, it can also be found in association with other abnormalities, including cervical spine abnormalities Klippel-Feil syndrome, Goldenhar syndrome, heterochromia, and congenital deafness. Differential diagnosis includes Duane-radial ray syndrome, acro-renal-ocular syndrome, Bosley-Salih-Alorainy syndrome, Townes-Brocks syndrome, Athabaskan brainstem dysgenesis-related disorders, Wildervanck syndrome, horizontal gaze palsy with progressive scoliosis, Moebius syndrome and congenital fibrosis of extraocular muscles. Management of the condition is mainly supportive and involves wearing spectacles or contact lenses to correct refractive errors. Surgical intervention to try and correct the face turn and to align the eyes in primary position can be attempted. However, surgery cannot correct the underlying innervations defect and, therefore, cannot remove the strabismus.

Molecular Genetics

Duane retraction syndrome usually presents sometimes as a sporadic disease. However, in about 10% of the cases, it presents in a familial fashion, with an autosomal dominant mode of inheritance. There is incomplete penetrance with variable expressivity. Defects in two separate loci have been implicated in the pathogenesis of this condition. The first is a locus on chromosome 8q. However, no candidate genes in this locus have so far been identified. The second locus identified was on chromosome 2q. Mutations in the CHN1 gene on this locus have been identified in families with isolated DRS.

Epidemiology in the Arab World

Saudi Arabia

Khan et al. (2007) undertook a retrospective analysis of all cases diagnosed with DRS at the



King Khaled Eye Hospital between 1982 and 2004. Of the 404 medical records studied, 45% were males, while 55% were females. There were more unilateral cases (86%) than bilateral, and among the unilateral cases, more cases had their left eyes affected (79%) than right eyes. Type I DRS was most common (78%), followed by Type III (19%) and Type I (4%). A family history of strabismus was seen in 13% of the cases, and 12% of the patients had other congenital anomalies in addition to DRS. Although there was a larger proportion of females compared to males in among the unilateral cases, it was the opposite in the bilateral cases. The overall features of DRS in Saudi Arabia were found to be similar to those reported from other parts of the world.

References

Khan AO, Oystreck DT, Wilken K, Akbar F. Duane retraction syndrome on the Arabian Peninsula. *Strabismus*. 2007; 15(4):205-8. PMID: 18058357 [FT]

Related CTGA Records

N/A

External Links

<http://emedicine.medscape.com/article/1198559-overview>

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

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=233

Contributors

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