



GOLDENHAR SYNDROME WITH BILATERAL DUANE RETRACTION SYNDROME: A RARE CASE

Wasel Shgeirat MD^{1*} and Mohammad Abu-Ain²

¹Oculoplastic Surgeon and Strabismologist.

²Consultant Ophthalmic Surgeon.



*Corresponding Author: Wasel Shgeirat
Oculoplastic Surgeon and Strabismologist.

Article Received on 06/11/2024

Article Revised on 26/11/2024

Article Accepted on 16/12/2024

ABSTRACT

Goldenhar syndrome is a rare congenital condition characterized by abnormal development of the eye, ear, heart, kidneys and spine. It is also called Oculo -Auriculo -Vertebral dysplasia. Duane retraction syndrome (DRS) is a congenital strabismus syndrome with abnormalities in horizontal eye movements, narrowing of the palpebral fissure and globe retraction +_ up and down shooting of the eye. Both syndromes are of congenital origin, and the association is rarely reported. We report a case of 17-year-old female patient who presented with hemifacial atrophy, facial microsomia, preauricular skin tags small ear and hearing loss with Duane features.

KEYWORDS: Goldenhar Syndrome, Duane retraction syndrome.

INTRODUCTION

Goldenhar syndrome (GS) is a congenital disorder affecting the structures of the face, especially the ones derived from the 1st and 2nd branchial arches.^[1]

Features of GS include

Hemifacial atrophy, epibulbar dermoids, preauricular skin tags and spinal cord defects.

Thus, GS is also known as^[2]

Oculoauriculocervical.
Facioauriculovertebral dysplasia.
First and 2nd branchial arches syndrome.
Unilateral mandibulofacial dysostosis.

Duane Retraction Syndrome(DRS)^[3]

Is a congenital strabismus disorder characterized by horizontal duction abnormalities, globe retraction and narrowing of the palpebral fissure on attempted adduction with up / down shooting sometimes.

Types

DRS1: abduction is affected 75%
DRS2: adduction is affected 5-10%
DRS3: both abduction and adduction affected 10-20 %

The exact cause is unknown, some theories suggest the absence of the 6th nerve nucleus with abnormal innervation of the LR by branches of the 3rd nerve.^[4]

Here we present a 17-year-old female with a rare case of Goldenhar Syndrome and bilateral Duane Retraction Syndrome association.

CASE REPORT

A 17-year-old female, born via full-term cesarean section, has a history of abnormal facial features (figures 1,2,3), hearing loss that treated first by hearing aids then cochlear implant was received (figure 4) and ventricular septal defect that was closed spontaneously,. strabismus: bilateral abduction deficit (figure 6), congenital hypothyroidism for which she is on thyroxine since one year of age.

she underwent squint surgery for esotropia with bilateral medial rectus recession (5 mm) and removal of ear skin tags. She has been regularly followed since then. On her most recent examination, her aided visual acuity was 6/6 in both eyes, with normal anterior and posterior segments. Refraction under cycloplegia (RUC) showed -4.00/-2.50×180 in the right eye and -3.75/-1.50×15 in the left eye, and she wears -4.00/-2.50×180 and -3.50/-1.25×10, respectively.

MRI revealed partial agenesis of the anterior falx cerebri with interdigitations between the medial aspects of the frontal lobes.

Cervical x-ray showed fused c3-c4 vertebral bodies with butterfly vertebrae and cervical scoliosis (figures 4,5)



Figure 1: Goldenhar Syndrome: RT Side Facial Microsomia.



Figure 3: Cochlear Implant and Fused Cervical Vertebrae.



Figure 2: The Site of Preauricular Skin Tags (Were Removed Already).



Figure 4: Fused Cervical Vertebrae and Cervical Scoliosis.



Figure 6: 9 Cardinal Positions of Gaze Showing Bilateral Abduction Deficit.

DISCUSSION

Goldenhar Syndrome (GS) primarily affects structures derived from the 1st and 2nd branchial arches, presenting unilaterally in 85% of cases and bilaterally in 15%, with the right side more frequently involved. The syndrome has a prevalence of 1 in 3,500–7,000 live births and a male-to-female ratio of approximately 3:2. While most cases are sporadic, autosomal dominant inheritance has been reported. Common anomalies include ocular involvement (60%), vertebral defects (40%), and ear abnormalities (40%). Duane Retraction Syndrome (DRS) is an unusual but recognized association, occurring in 5–10% of cases. Most GS cases (~70%) are isolated, but 30% are associated with other anomalies. This case is notable for two rare features: the association of GS with DRS and the bilateral involvement of Duane Syndrome, an exceedingly uncommon.^{[5] [6] [7]}

*Patient consent was taken

1. <https://pmc.ncbi.nlm.nih.gov/articles/PMC6117527/>
2. <https://rarediseases.org/rare-diseases/oculo-auriculo-vertebral-spectrum/#synonyms>
3. https://eyewiki.org/Duane_Retraction_Syndrome
4. <https://emedicine.medscape.com/article/1198559-overview?form=fpf>
5. <https://www.herdin.ph/index.php?view=research&cid=69878>
6. <https://pubmed.ncbi.nlm.nih.gov/23279771/>
7. https://www.researchgate.net/publication/234031229_Goldenhar_Syndrome_in_Association_with_Duane_Syndrome