# Goldenhar Syndrome in Association with Duane Syndrome

Shrestha UD, Adhikari S
Pediatric Ophthalmology Unit, Tilganga Institute of Ophthalmology, Kathmandu, Nepal.

## ABSTRACT

Goldenhar syndrome (GHS) is also known as Oculo-Auriculo-Vertebral (OAV) syndrome or Branchial arch syndrome. Duane retraction syndrome (DRS) is a congenital disorder of ocular motility characterized by limited abduction, adduction or both. It is unilateral in 80% of cases. The important and interesting part of this eight months old child is presence of GHS with DRS. She has bilateral involvement, which is seen in only 5-8% of GHS, as compared to high incidence of unilateral involvement. This child also had refractive error of + 6.00/ - 1.5 * 180. At four year of age her vision with glass was 6/9. Children with GHS and DRS should have early eye examination done to treat the problem of refractive error.

**Keywords:** *Duane retraction syndrome; goldenhar syndrome, refractive error*

## INTRODUCTION

Goldenhar syndrome (GHS) is also known as Oculo-Auriculo-Vertebral (OAV) syndrome or Branchial arch syndrome. The pathognomonic triad includes presence of dermolipoma, craniofacial microsomia and vertebral skeletal anomalies.[1,2] The anomalies occurs at all levels within the spine. A wide range of anomalies may be present, including butterfly vertebrae; hemi vertebrae, which produce secondary scoliosis; kyphosis; and rib anomalies. These anomalies cannot be predicted from the severity of the facial malformation.[3]

It is associated with anomalous development of the first branchial arch and second branchial arch.[4] Only in 10% of cases GHS is bilateral. GHS was documented in 1952 by Maurice Goldenhar (1924-2001).[5] Patient with oculo-auriculo-vertebral spectrum have bilateral cleft lip and palate, macrostomia, malformed pinna, and unilateral hypoplasia of the zygomatic arch and mandible, which result in an asymmetric facial appearance characteristic of hemifacial microsomia.[6]

Other associated malformations include strabismus, orbital hypoplasia, upper lid colobomas, nasolacrimal duct obstruction, iris coloboma, persistent pupillary membrane, and hypoplasia of the pinna, fusion of cervical vertebrae, scoliosis, renal agenesis and multicystic kidney.

## CASE REPORT

Eight months old girl child presented with some elevation on the white part of eye since birth. She had restricted eye movement on the left side since birth. On examination, the child fixated and followed the light. She had esotropia of about 15 degree on Hirshberg light reflex test (Figure 1).

Correspondence:
Dr. Ujjowala Devi Shrestha
Pediatric Ophthalmology Unit
Tilganga Institute of Ophthalmology
National Academy of Medical Sciences, NAMS
Phone: 4493775
Email: ujjowala@gmail.com

Figure 1. Esotropia in primary position

Both on dextroversion and levoversion she had the overshooting with the inferior oblique overaction in both the eyes (Figure 2 and 3).

Figure 2. Dextroversion: Inferior oblique overaction of the left eye

Figure 3. Levoversion: Inferior oblique overaction of the right eye

On extra-ocular motility examination, she had restricted abduction with the classic retraction of the globe and narrowing of the palpebral fissure in adduction (Figure 4).

Figure 4. Figure 4. Abduction deficit in the left eye

Anterior segment examination revealed the lipo dermoid since birth on the right eye (Figure 5).

Figure 5. Lipo dermoid of right eye

Cornea was clear. Pupil was round regular and reacting to light. On cycloplegic refraction, her refractive error was mixed astigmatism of + 6.00/ - 1.5 * 180. Fundal examination showed normal findings. She had pre auricular tag in both the ears (Figure 6 and 7).

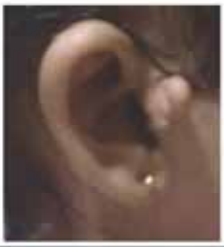

Figure 6. Preauricular tag of right ear

Figure 7. Preauricular tag of left ear

There was no other systemic associations or spinal anomalies.

This child was diagnosed as GHS with DRS. She was on regular follow up at every 4-6 months interval. At 4 years of age, her best corrected visual acuity was 6/9 in both the eyes with +4.00/-0.5 *180 glasses in both the eyes (Figure 8).

Figure 8. Child with glass

## DISCUSSION

The oculo-auriculo-vertebral dysplasia, as described by Goldenhar, is a disease complex of structures developed from the first and second branchial arch. The interesting part of this case is bilateral involvement, seen in only 5-8% of Goldenhar syndrome, as compared to high incidence of unilateral involvement.[1] The other important finding of this case is its association with Grade I DRS. DRS existing with GHS have been reported earlier.[7] The pathological basis of ocular motility disorders with GHS remains unknown. However, the aplasia of abducens nerve and nucleus has been studied.

Duane retraction syndrome is a congenital disorder of ocular motility characterized by limited abduction, adduction or both. It is unilateral in 80% of cases. DRS have three presentations. Type 1 occurs in 85% of cases and predominantly in females (60%). It presents with abduction palsy with globe retraction on attempted

adduction. Type II DRS occurs in 14% of cases, and presents as an adduction palsy, globe retraction on attempted adduction and intact abduction. Type-III DRS comprises 1% of cases; both abduction and adduction limitations with any form of horizontal strabismus characterize this type. All cases may present with the "tether" phenomenon, with over-elevation or over-depression in adduction.

In 70 percent of DRS cases, this is the only disorder the individual has. However, thirty percent of the cases have other conditions and syndromes in association with DRS. These include malformation of the skeleton, ears, eyes, kidneys and nervous system, as well as:

- Okihiro's syndrome, an association of DRS with forearm malformation and hearing loss,
- Wildervanck syndrome, fusion of neck vertebrae and hearing loss,
- Holt-Oram syndrome, abnormalities of the upper limbs and heart,
- Morning Glory syndrome, abnormalities of the optic disc or "blind spot", and
- Goldenhar syndrome, malformation of the jaw, cheek and ear, usually on one side of the face.[8]

Thirty percent of cases have limb abnormalities, cardiac abnormalities, neurosensory deafness, Klippel-Feil syndrome, congenital gustolacrimal reflex and Wildervanck association. Children with GHS and DRS may have refractive error. Amblyopia and absent binocular stereo vision affected one in five patients with DRS.[9] Unlike unilateral DRS, bilateral DRS may be more common in males and associated with a higher prevalence of strabismus in primary gaze position. The prevalences of amblyopia, positive strabismus family history, and associated congenital abnormalities in this series of bilateral cases is similar to the reported prevalence.[10] Early eye examination help to correct the problem of refractive error. Hence, early referral for eye examination should be done for children with GHS and DRS.

## REFERENCES


1. Harley RD. Paediatric Ophthalmology. 2nd ed. Philadelphia : W.B. Saunders Company; 1983.
2. Feingold M, Gabis S. Ocular abnormality associated with 1st and lied arch syndrome. Survey Ophthal. 1968;14:30.
3. Anderson PJ, David DJ. Spinal anomalies in Goldenhar syndrome. Cleft Palate Craniofac J. 2005 Sep;42,5:477-80.
4. Touliatou V, Fryssira H, Mavrou A, Kanavakis E, Kitsiou-Tzeli S. Clinical manifestations in 17 Greek patients with Goldenhar syndrome. Genet Couns. 2006; 17,3:359–70.
5. Vinay C, Reddy RS, Uloopi KS, Madhuri V, Sekhar RC. Craniofacial features in Goldenhar syndrome. J Indian Soc Pedod Prev Dent. 2009 Apr-Jun; 27,2:121–4.
6. Suda N, Watanabe M, Ohyama K. A case of oculo-auriculo-vertebral spectrum with long-term and comprehensive management. World J Orthod. 2006; 7,3:299-304.
7. Mansour AM, Wang F, Henkind P, et al. Ocular Findings in Goldenhar Syndrome. Am J Ophthalmol. 1985;100:555-9.
8. Javed EA. Familial Duane's Syndrome. Professional Med J. Sep 2010;17,3:522-5.
9. Chua B, Johnson K, Donaldson C, Martin F. Management of Duane retraction syndrome. J Pediatr Ophthalmol Strabismus. 2005 Jan-Feb; 42,1:13-7; quiz 45-6.
10. Khan AO, Oystreck D. Clinical characteristics of bilateral Duane syndrome. J AAPOS. 2006 Jun; 10,3:198-201.