

Brief Communication

Ocular Manifestations of Goldenhars syndrome- A case series

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Abstract

Goldenhars syndrome also known as facio auriculo vertebral dysplasia or first and second arch branchial syndrome has a prevalence of 1:3500 births to 1: 7500 births with male predisposition. We hereby present a case series of children presenting with Goldenhars syndrome.

Brief communication

Goldenhars syndrome also known as facio auriculo vertebral dysplasia or first and second arch branchial syndrome is characterized by triad of anomalies comprising of ocular epibulbar dermoid, accessory auricular appendages and aural fistula. (Stanojevic, 2000; Pridijian, 1995)

Ocular presentation consists of unilateral or bilateral epibulbar dermoids in 35% of cases. These may appear as yellowish or pinkish ovoid masses which vary from pin sized epidermoid to 8- 10 mm swelling with predisposition for inferotemporal quadrant of limbus. They are also associated with coloboma of eyelids or

iris or choroid. Other ocular features include strabismus, epicanthal folds, anophthalmia, anomalies of lacrimal drainage system. (Jakobiec, 2016) Other systemic findings are preauricular tag, facial asymmetry due to cleft lip or palate and vertebral dysplasia. All these features predispose a baby to difficulty in breathing, hearing and visual impairment.

We present a series of children with Goldenhar syndrome presenting with various systemic and ocular anomalies.

Case 1

A six month old baby boy presented to the ophthalmology department of a pediatric tertiary care hospital with complaints of something projecting out from both the eyes. On examination, there was an epibulbar dermoid with a cutaneous horn of size 10 mm x 3mm projecting out from both the eyes (OU). The child was fixing and following at torch light. Rest of the examination of anterior and posterior segment was unremarkable. Systemic examination revealed two lipomas measuring 2 cm X 1.5 cm protruding out from his scalp and 1 near his left ear. Rest of the systemic examination was within normal limits. The child was advised epibulbar dermoid excision after 1 year of age. (Figure 1)

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Case 2

A 10 year old boy presented with inward deviation of eyes since birth. There was no history of using glasses. Child was operated for duodenal atresia in childhood. On ocular examination, Best corrected visual acuity (BCVA) was 6/12 in right eye and 6/6 in left eye. There was esotropia with limited abduction in both eyes. It was associated with narrowing of palpebral fissure on adduction and widening of palpebral fissure on abduction suggestive of bilateral Duane's retraction syndrome. Upshoot was present in both eyes. Krimsky test measured 60 PD ET. Lipodermoid was present in both eyes inferotemporally. Preauricular tags were present on right side. Cycloplegic refraction revealed hypermetropic astigmatism in both eyes with absence of anisometropia. Child underwent preauricular tag removal for cosmetic purpose and was prescribed glasses. Child was advised occlusion therapy for right eye amblyopia. Surgical management was performed for primary gaze esotropia in the form of bilateral medial rectus recession under

General anesthesia. No other systemic anomaly noted. (Figure 2)

Case 3

A 6 year old male child presented for routine ophthalmic examination. Old records revealed of surgical correction of cleft lip and cleft palate. Child was having congenital nasolacrimal duct obstruction which was managed successfully by sac massage. On examination child had facial asymmetry with Preauricular tag on right side. Ocular examination revealed BCVA of 6/60 OD, 6/6 OS. OD revealed microphthalmos with microcornea. Lipodermoid was present in right eye inferotemporally associated with bulging of lower lid. Ophthalmic examination of OS was normal. Child was treated with occlusion therapy in OS. No surgical correction was done for dermoid as parents were not willing. (Figure 3)

Goldenhar syndrome can present with variety of ocular features. Management depends on presentations and visual potential.



Figure 1: 6 month old male, Anterior segment examination of both eyes (a,b) showing epibulbar dermoid with cutaneous horn. Facial examination showing lipomas protruding from scalp (c) and lipoma near left ear (d).



Figure 2: Case 2, 10 year old male, Anterior segment examination of both eyes showing (a, b) esotropia with palpebral fissure changes (c) upshoot in left eye (d,e) lipodermoid inferotemporally. Facial examination revealed (f) Preauricular tag near right ear



Figure 3: Case 3, 6 year old male, Facial examination showing (a) repaired cleft lip (b) preauricular tag. Anterior segment examination of both eyes showing (c,d) lipodermoid inferotemporally. (e) Oral examination revealed cleft palate. Anterior segment examination of right eye showing (f) microphthalmos with microcornea

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