

Ocular Manifestations Of Goldenhar Syndrome; At A Glance

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Abstract Goldenhar syndrome (oculo-auriculo-vertebral dysplasia, OAVS) is a rare, congenital disease with an incidence of 1:3500 and 1:5600. It occurs due to abnormal development of the first and second branchial arches. It has a multifactorial etiopathogenesis. The typical features of Goldenhar syndrome include ocular anomalies - epibulbar dermoids, microphthalmia and coloboma, auricular features such as preauricular tags, hearing loss, micrognathia, and vertebral anomalies such as scoliosis or hemivertebrae.

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Introduction

Goldenhar syndrome (oculo-auriculo-vertebral dysplasia, OAVS) is a rare, congenital disease occurring due to abnormal development of the first and second branchial arches.¹ It has a multifactorial etiopathogenesis and dependent on genetic and environmental factors.² It is associated with constellation of anomalies including ocular, craniofacial (cleft lip / palate, mandibular hypoplasia, etc) (Figure 1), auricular (preauricular appendages / fistulas, ear asymmetry, microtia,etc), (Figure 2) and skeletal (vertebral defects, etc) deformities.³⁻⁶

Ocular Manifestations

The classic ocular features of this syndrome include microphthalmia, epibulbar dermoids, lipodermoids(Figure3) and coloboma. However, there are many additional ocular signs that can be found in a patient of Goldenhar syndrome associated with systemic features. (Table 1)



Figure 1: 12 year old child with Goldenhar syndrome; showing mandibular hypoplasia

Figure 2: Auricular features showing microtia with external auditory canal atresia and a pre-auricular skin tag (blue arrow) of right ear



Figure 3: Ocular findings showing dermolipoma in right eye (red arrow) & Limbal dermoid in left eye encroaching visual axis (yellow arrow).

Table 1: Common ocular manifestations of Goldenhar syndrome

	Ocular manifestations
Eyeball, Eyelid	Eyelid coloboma, blepharoptosis, microphthalmos, anophthalmos, cryptophthalmos. ⁷⁻⁹ Associated ocular complications with eyelid coloboma: Exposure keratopathy, corneal ulceration, corneal perforation, corneal leukoma, amblyopia
Lacrima system	Congenital dacryostenosis, lacrimal drainage obstruction, lacrimal fistula, lacrimal sinus dimple. ¹⁰
Strabismus	Esotropia followed by exotropia, congenital ophthalmoplegia, Duane's retraction syndrome. ¹¹
Conjunctiva	Dermolipomas, caruncular abnormalities(dysplastic and/ or bilobed caruncles, ectopic caruncles) ¹²
Cornea, limbus	Limbal dermoids, microcornea, corneal anesthesia, neuroparalytic keratitis. ¹³ Associated ocular complications with limbal dermoids: corneal astigmatism, anisometric amblyopia, visual axis obscuration, ocular surface abnormalities, dellens, inadequate lid closure. ¹⁴
Iris /choroid	Coloboma ¹⁵
Lens	Cataract(polar cataract)
Optic nerve/disc	tilted optic disc, optic nerve hypoplasia, disc coloboma
Retina	Vessel tortuosity, retinal hypopigmentation, hypoplastic macula Exudative Vitelliform Maculopathy has also been found associated ¹⁶

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