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Case Series

Ophthalmology Section

Isolated Oculo-auricular Manifestations in Goldenhar Syndrome: A Case Series of Three Paediatric Patients

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ABSTRACT

Goldenhar Syndrome (GS), also known as the Oculo-Auriculo-Vertebral Spectrum (OAVS), is a rare congenital disorder characterised by variable craniofacial and ocular anomalies due to aberrant development of the first and second branchial arches. This case series describes three paediatric patients who exhibited diverse phenotypic features of the syndrome, highlighting its clinical heterogeneity and the importance of individualised management. All three patients presented with epibulbar dermoids since birth, an ocular hallmark of GS. The first patient, a 12-year-old female, had a limbal dermoid, medial canthal swelling suggestive of nasolacrimal duct obstruction, a symblepharon, and microtia with preauricular tags. She was advised dacryocystorhinostomy and excision of the dermoid but was lost to follow-up. The second patient, a 15-year-old had a reddish limbal mass extending to the lateral canthus and preauricular tags, and a cosmetic symblepharon release was advised. The third patient, a 15-year-old had a non-progressive limbal dermoid with no significant adnexal involvement and was managed conservatively. None of the patients had associated spinal, cardiac, or neurological anomalies on systemic evaluation. This case series reinforces the phenotypic variability of GS and underscores the importance of early ophthalmic diagnosis, systemic screening, and a multidisciplinary approach to optimise functional and cosmetic outcomes in affected children.

Keywords: Congenital anomaly syndromes, Craniofacial anomalies ear malformations, Ocular dermoids

INTRODUCTION

Goldenhar Syndrome (GS), also known as the Oculo-Auriculo-Vertebral Spectrum (OAVS), is a developmental disorder involving tissues derived from the first and second branchial arches. First described by Goldenhar in 1952, the syndrome is classically defined by a triad of hemifacial microsomia, auricular anomalies, and epibulbar dermoids. It may also involve vertebral, cardiac, renal, and central nervous system abnormalities [1,2].

The estimated incidence ranges from 1 in 3,500 to 1 in 45,000 live births. GS shows a male predominance and is usually unilateral. Although its aetiology remains largely idiopathic, both genetic and environmental factors have been implicated [3]. Early recognition is critical due to potential multi system involvement and the need for coordinated care across specialties [4]. The syndrome is classically defined by a triad of hemifacial microsomia, auricular anomalies, and epibulbar dermoids [5].

Here, we present a case series of three children with GS, each illustrating different levels of systemic and ocular involvement.

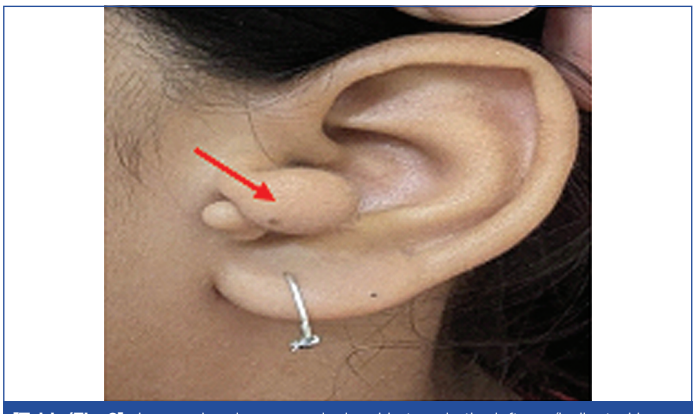
CASE SERIES

Case 1

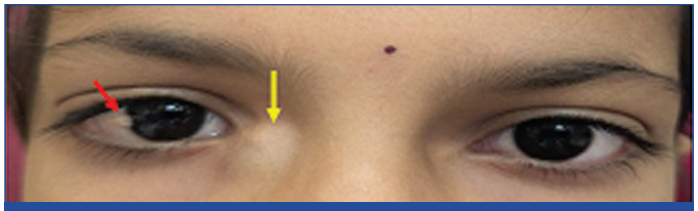
A 12-year-old female, born of non-consanguineous marriage, presented to the ophthalmology outpatient clinic with a chief complaint of a mass near the right eye since birth. The mass was noted by her parents when she was born. It was painless, nonprogressive, and not associated with discharge. The child was born full term via an uneventful vaginal delivery, had a normal birth weight, did not require neonatal intensive care, and was vaccinated according to the immunization schedule. Developmental milestones were age-appropriate, and there were no known cognitive delays.

On general physical examination, the patient had microtia of the right ear [Table/Fig-1] and preauricular skin tags on the left ear [Table/Fig-2]. Spinal examination revealed no abnormalities, and there was no evidence of a high-arched palate. On ophthalmic evaluation, Best Corrected Visual Acuity (BCVA) was 6/6 in both eyes, with a refractive error of -1.00 diopter cylinder at axis 180° in the right eye. On torchlight examination, a firm, non-tender, globular mass measuring approximately 1×1 cm was observed medial to the right medial canthus [Table/Fig-3]. The overlying surface was smooth with a greenish hue, without redness, tenderness, discharge, pustules,

[Table/Fig-1]: Image showing microtia of the right ear (indicated by red arrow).

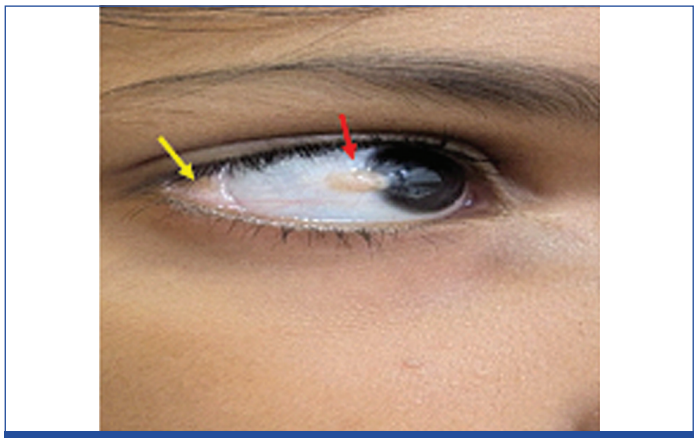


[Table/Fig-2]: Image showing pre auricular skin tags in the left ear (indicated by red arrow).



[Table/Fig-3]: Image showing a mass near the right medial canthus due to lower punctum stenosis (indicated by yellow arrow) and limbal dermoid (indicated by red arrow).

or superficial vascularisation. Extraocular movements were full and unrestricted in all gazes, and the remainder of the ocular adnexa was normal. On slit-lamp examination, a pale, elevated, avascular, hemispherical mass approximately 0.5 mm in diameter was noted at the 9 o'clock position on the temporal limbus of the right eye, consistent with a limbal dermoid. Additionally, a conjunctival fold near the lateral canthus was noted, likely representing a symblepharon [Table/Fig-4]. The lower canalicular punctum was absent in the right eye. The rest of the anterior segment examination was unremarkable. Dilated fundus examination of both eyes was normal. A cardiac evaluation by 2D echocardiography, performed to screen for associated congenital anomalies, was normal. No renal anomalies were reported. The limbal lesion presented a clinical appearance consistent with a grade 1 limbal dermoid (Mansour AM et al.,), but differential diagnosis included epibulbar dermolipoma, conjunctival papilloma, and choristoma variants [6]. The medial canthal swelling was suggestive of congenital nasolacrimal duct obstruction, though dacryocystocele and medial dermoid cyst were also considered. The presence of preauricular skin tags and microtia pointed toward a syndromic cause, most consistent with GS (OAV spectrum). Infective or inflammatory causes for symblepharon were ruled out based on congenital onset and lack of inflammatory signs.



[Table/Fig-4]: Image showing a limbal dermoid (indicated by red arrow) with symblepharon (indicated by yellow arrow) in the right eye.

The patient was advised to undergo dacryocystorhinostomy to address the problem of lacrimal drainage abnormality. However, she was lost to follow-up, and further management could not be pursued.

Case 2

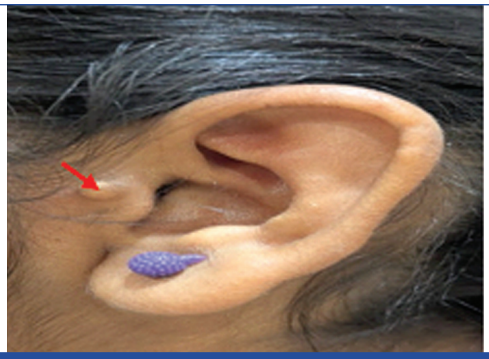
A 15-year-old female, accompanied by the parents who were also the informants, presented to the ophthalmology outpatient department with chief complaints of reddish discolouration in the left eye. According to the parents, the lesion had been present since birth, was painless and non-progressive in size. The patient was born of nonconsanguineous marriage at full term via an uncomplicated vaginal delivery, with a normal birth weight and no history of admission to the neonatal intensive care unit. Her immunisations were up to date. Developmental milestones were age-appropriate, and no intellectual disability was noted. On general physical examination, a preauricular skin tag was observed on the left-side[Table/Fig-5], while the right ear appeared anatomically normal. There were no signs of spinal deformity, and the palate was of normal morphology, with no evidence of a high-arched palate. Upon ophthalmic evaluation, she had a BCVA of 6/6 in both eyes. Extraocular movements were full and unrestricted in all gazes. Examination of the ocular adnexa was unremarkable. On slit-lamp biomicroscopy, the left eye demonstrated a reddish conjunctival mass on the temporal aspect, extending from the limbus to the lateral canthus, likely to be a symblepharon [Table/Fig-6]. The rest of the anterior segment examination was otherwise normal in both eyes. The dilated fundus examination of both eyes was normal. A detailed systemic evaluation, including cardiac assessment, was performed and found to be within normal limits. The differential diagnosis for the temporal conjunctival mass consistent with symblepharon included conjunctival papilloma, vernal limbitis, and post-inflammatory conjunctival fibrosis. However, the lesion's congenital onset, stability, and association with auricular skin tags were more suggestive of choristomatous origin, consistent with GS. There was no history of trauma, chemical injury, or ocular surgery to suggest an acquired symblepharon. In view of cosmetic concern, the patient was advised surgical symblepharon release and was appropriately counselled regarding the possibility of recurrence post-procedure. The patient did not opt for surgery due to the possibility of recurrence.

[Table/Fig-5]: Image showing pre auricular skin tag of the left ear (indicated by red arrow).

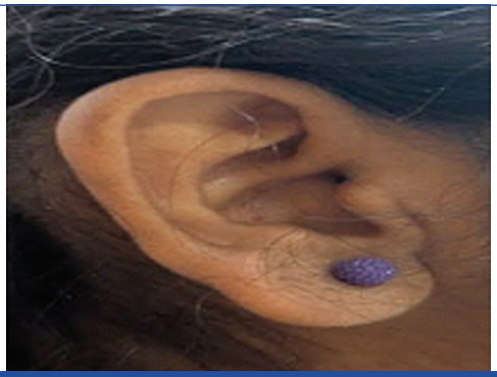
[Table/Fig-6]: Image showing symblepharon in the left eye (indicated by red arrow).

Case 3

A 15-year-old female, accompanied by her mother who was the informant, presented to the ophthalmology outpatient department with a mass in the left eye. The lesion had been present since birth and was described as painless but gradually increasing in size over the years. She was born at full term to non-consanguineous parents, with a normal birth weight and no requirement for neonatal intensive care. She was immunised to date. Developmental milestones were age-appropriate, and there was no evidence of cognitive impairment. General physical examination revealed a preauricular skin tag on the left-side [Table/Fig-7], whereas the right ear was normal [Table/Fig-8]. There were no craniofacial abnormalities, and examination of the spine did not reveal any vertebral anomalies.



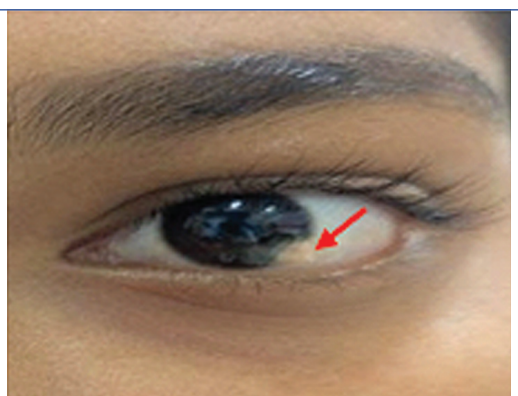
[Table/Fig-7]: Image showing pre auricular skin tag of the left ear (indicated by red arrow).



[Table/Fig-8]: Normal right ear.

The patient was otherwise systemically stable, with no additional dysmorphic features noted.

Ophthalmological evaluation revealed BCVA of 6/6 in both eyes. Extraocular movements were full and unrestricted in all directions of gaze. The ocular adnexa were unremarkable bilaterally. On slit-lamp examination, the anterior segment of the right eye was within normal limits. The left eye showed a solitary, well-defined, globular mass located at the limbus. It had a smooth, uniform surface and lacked associated vascularisation-clinically suggestive of a limbal dermoid [Table/Fig-9]. The rest of the anterior segment examination of the left eye was unremarkable. Dilated fundus examination was normal in both eyes. A comprehensive cardiac evaluation demonstrated no abnormalities. The limbal mass was most consistent with a grade 1 epibulbar dermoid, but differentials included conjunctival nevus, dermolipoma, and other ocular surface choristomas. The lack of pigmentation, vascularisation, and systemic features ruled out neoplastic causes. The coexisting preauricular skin tag and normal systemic examination further supported a mild form of GS (OAV spectrum), even in the absence of other craniofacial anomalies.



[Table/Fig-9]: Image showing limbal dermoid in the left eye (indicated by red arrow).

As the lesion was not visually significant and the patient reported no discomfort, she was counselled regarding the benign nature of the condition. No active ophthalmic intervention was deemed necessary at present, and the patient was advised regular follow-up for monitoring.

DISCUSSION

GS, first described by Maurice Goldenhar in 1952 [1], is a congenital craniofacial disorder within the OAV spectrum. It results from abnormal development of the first and second branchial arches and typically manifests with epibulbar dermoids, auricular anomalies, and vertebral defects. The incidence of GS ranges from 1 in 3,500 to 1 in 5,600 live births, with a male predominance and more frequent right-sided involvement [7].

The case series presented three paediatric patients who had varying degrees of ocular and auricular anomalies without vertebral, cardiac, or CNS involvement. This supports the observation that GS can exist on a clinical spectrum, with isolated oculo-auricular presentations representing a milder form [4,5]. The most common ocular finding in our cases was the presence of limbal dermoids, which are a hallmark of GS. These findings are consistent with previous reports from Indian literature, where conjunctival or corneal dermoids were also reported as prominent manifestations [7,8]. Case 1 and Case 2 demonstrated a symblepharon, which is an uncommon finding in GS but has been mentioned in case reports involving more extensive ocular surface anomalies [7].

Auricular anomalies such as preauricular tags and microtia were noted in all three patients. These are among the most common features of GS and may occur in up to 85% of affected individuals [2,4]. None of our patients exhibited vertebral, renal, or cardiac anomalies, though these are known to occur in 30-50% of cases [9]. The phenotypic heterogeneity of GS has been well documented, with wide variability in the severity and combination of craniofacial and systemic manifestations [5,10].

Their absence in our series again suggests a milder phenotype. Management strategies in our patients were tailored to clinical severity. Observation was sufficient for minor ocular surface anomalies, while surgical excision was employed for symptomatic or visually significant dermoids. This approach aligns with current guidelines advocating for individualised, multidisciplinary care planning based on severity and functional impact [11]. Psychosocial and cosmetic concerns, especially in children approaching school age, were also taken into account in the decision-making process [12].

CONCLUSION(S)

This case series highlights the wide variability in how GS can present, even among patients of similar age. While all cases had ocular and auricular signs, none showed systemic involvement, suggesting a mild form of the syndrome. This experience shows that individualised treatment-whether surgical, cosmetic, or observational-can be effectively tailored to each child's needs. Early diagnosis and multidisciplinary assessment are essential for good long-term outcomes. Awareness of these subtle presentations can help avoid delayed or missed diagnosis.

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