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GOLDENHAR SYNDROME: A COMPREHENSIVE CASE REPORT AND REVIEW

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ABSTRACT Goldenhar Syndrome, also known as oculo-auriculo-vertebral (OAV) spectrum, is a rare congenital condition characterized by craniofacial anomalies, including facial asymmetry, ear abnormalities, and vertebral defects. This case report details an 8-year-old male patient, deaf and mute, who presented to the ophthalmology outpatient department with a corneal ulcer in his right eye and a limbal dermoid in his left eye. The patient had a history of cleft lip and cleft palate surgery in childhood and exhibited ear anomalies. This report aims to highlight the clinical presentation, diagnosis, and management of Goldenhar Syndrome in this patient.

KEYWORDS : Goldenhar Syndrome, oculo-auriculo-vertebral spectrum, craniofacial anomalies, corneal ulcer, limbal dermoid, congenital ear anomalies, cleft lip and palate.

INTRODUCTION

Goldenhar Syndrome is a complex congenital disorder first described by Maurice Goldenhar in 1952. It involves anomalies in structures derived from the first and second branchial arches. The syndrome manifests with a spectrum of craniofacial abnormalities, often including ocular, auricular, and vertebral malformations. The etiology of Goldenhar Syndrome is multifactorial, involving genetic and environmental factors. The incidence is estimated to be between 1 in 3,500 to 1 in 5,600 live births, with a higher prevalence in males.[1-3]

CASE REPORT

An 8-year-old male patient, deaf and mute, presented to the ophthalmology outpatient department with complaints of pain, redness, and decreased vision in his right eye for the past week. On examination, a corneal ulcer was noted in the right eye. Additionally, a limbal dermoid was observed in the left eye. The patient's medical history revealed he had undergone cleft lip and cleft palate surgery during early childhood. Examination of the ears showed structural anomalies consistent with microtia.

OPHTHALMIC EXAMINATION

- Right Eye: Visual acuity was significantly reduced due to the presence of a corneal ulcer. Slit-lamp examination revealed a large central corneal ulcer with stromal involvement.
- Left Eye: Visual acuity was within normal limits. A limbal dermoid was present at the inferotemporal limbus, not affecting the visual axis.

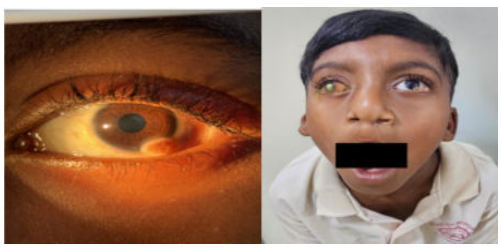

Fig 1a Showing slit lamp photo of limbal Dermoid in Left Eye. 1b Shownig Corneal Ulcer in Right Eye and Limbal Dermoid in Left eye.

SYSTEMIC EXAMINATION

- Craniofacial Anomalies: The patient had facial asymmetry, with the right side of the face appearing smaller than the left.
- Auricular Anomalies: Both ears exhibited microtia with preauricular skin tags.
- Previous Surgical Interventions: Scarring from previous cleft lip and palate surgery was evident.

DIAGNOSTIC WORKUP

- Hearing Assessment: Confirmed profound bilateral sensorineural hearing loss.
- Radiological Imaging: CT scan of the head and neck showed asymmetry of the facial bones and vertebral anomalies, particularly cervical vertebrae fusion.

MANAGEMENT AND FOLLOW-UP

The corneal ulcer in the right eye was treated with intensive topical antibiotics and cycloplegics. The limbal dermoid in the left eye was monitored, with surgical excision planned if it interfered with vision or caused discomfort. Regular follow-ups were scheduled to monitor ocular health and manage any recurrent infections or complications.

Fig 2a Showing Craniofacial Abnormalities. Fig 2b Showing Auricular Deformity.

DISCUSSION

Goldenhar Syndrome presents with a diverse array of clinical features, making diagnosis and management challenging. This syndrome's hallmark is the combination of craniofacial anomalies, which can vary widely in severity and presentation. The patient in this case exhibited significant ocular manifestations, including a corneal ulcer and a limbal dermoid, which are relatively uncommon but notable features of Goldenhar Syndrome.[4,5]

The presence of these ocular manifestations in conjunction with other craniofacial abnormalities such as facial asymmetry and microtia (small ears with preauricular skin tags) underscores the syndrome's complexity. The etiology involves disruptions in the development of the first and second branchial arches, leading to the characteristic abnormalities seen in affected individuals. Genetic studies have implicated several genes, although the precise genetic basis remains elusive. Additionally, environmental factors such as maternal diabetes and teratogenic exposures during pregnancy have been suggested as contributing factors.[6]

Management of Goldenhar Syndrome requires a multidisciplinary approach. In this case, the immediate concern was the treatment of the corneal ulcer, which was managed with intensive topical antibiotics and cycloplegics to reduce inflammation and prevent further damage. The limbal dermoid was monitored for potential surgical excision if it interfered with vision or caused discomfort.[7]

Early intervention, particularly for hearing and speech development, is crucial for improving the quality of life in affected individuals. This patient, being deaf and mute, likely faced significant communication

challenges, highlighting the importance of early and continuous audiological and speech therapy interventions.[8,9]

CONCLUSION

This case report underscores the importance of a comprehensive evaluation and management strategy for patients with Goldenhar Syndrome. The multidisciplinary approach, involving ophthalmologists, otolaryngologists, plastic surgeons, and audiologists, is essential for addressing the complex and varied needs of these patients. Further research into the genetic and environmental factors contributing to Goldenhar Syndrome is necessary to enhance diagnostic accuracy and develop more effective therapeutic strategies. Understanding the full spectrum of clinical manifestations and the underlying pathophysiology will facilitate better patient outcomes and improved quality of life for those affected by this rare congenital condition.

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