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Ophthalomology

GOLDENHAR SYNDROME ASSOCIATED WITH POSTAXIAL POLYDACTYLY: A RARE CASE REPORT

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ABSTRACT **Background:** Goldenhar syndrome, also known as Oculo-Auriculo-Vertebral Spectrum (OAVS), is a rare congenital developmental disorder involving structures derived from the first and second branchial arches. It is classically characterized by ocular dermoids, auricular anomalies, facial asymmetry, and vertebral defects. **Case Presentation:** A 6-year-old female child presented with a congenital yellowish-white mass over the left eye. Ocular examination revealed an inferotemporal limbal dermoid with mild left upper lid ptosis. Systemic examination demonstrated facial asymmetry, left hemifacial hypoplasia, left preauricular appendage, and postaxial polydactyly. Detailed pediatric evaluation, chest radiography, echocardiography, abdominal ultrasonography, and systemic examination revealed no associated cardiac, renal, neurological, or respiratory abnormalities. Based on the characteristic ocular and craniofacial findings, a diagnosis of Goldenhar syndrome was established. **Conclusion:** This case highlights the importance of comprehensive systemic evaluation in children presenting with limbal dermoids. The association of Goldenhar syndrome with postaxial polydactyly is uncommon and broadens the phenotypic spectrum of the disorder.

KEYWORDS : Goldenhar Syndrome; Oculo-Auriculo-Vertebral Spectrum; Limbal Dermoid; Preauricular Tag; Hemifacial Microsomia; Polydactyly.

INTRODUCTION

Goldenhar syndrome, first described by Maurice Goldenhar in 1952, is a rare congenital anomaly involving abnormal development of structures derived from the first and second branchial arches.[1] It is currently considered part of the Oculo-Auriculo-Vertebral Spectrum (OAVS).[2]

The estimated incidence ranges from 1 in 3,500 to 1 in 25,000 live births, with a slight male predominance.[3] Typical manifestations include epibulbar dermoids, preauricular appendages, microtia, facial asymmetry, mandibular hypoplasia, and vertebral abnormalities.[4]

Ocular findings are among the most characteristic features and frequently constitute the presenting complaint. Epibulbar dermoids represent the most common ophthalmic manifestation and may induce significant corneal astigmatism resulting in amblyopia.[5]

We report a case of Goldenhar syndrome presenting with unilateral limbal dermoid, facial asymmetry, preauricular appendage, and postaxial polydactyly without systemic involvement.

Case Report

A 6-year-old female child presented to the Ophthalmology Outpatient Department with a congenital yellowish-white mass over the left eye present since birth. There was no history of ocular trauma, pain, redness, watering, discharge, or previous ocular surgery.

Ocular Examination

Best-corrected visual acuity was 6/6 in the right eye and 6/12 in the left eye.

Slit-lamp examination of the left eye revealed a well-circumscribed yellowish limbal dermoid located at the inferotemporal limbus involving the superficial cornea and adjacent conjunctiva. Mild left upper lid ptosis was present.

The anterior chamber was quiet. Iris, lens, and posterior segment findings were within normal limits in both eyes.

Systemic Examination

General examination revealed:

- Facial asymmetry predominantly involving the left side
- Left hemifacial hypoplasia
- Left preauricular appendage (accessory tragus)
- Postaxial polydactyly of the hand

No clinically apparent vertebral abnormality was noted.

Systemic Evaluation

The patient underwent detailed pediatric evaluation.

The following investigations were within normal limits:

- Chest radiograph
- Two-dimensional echocardiography
- Ultrasonography of abdomen and kidneys
- Cardiovascular examination
- Central nervous system examination
- Respiratory system examination

No associated cardiac, renal, respiratory, or neurological abnormalities were identified.

Based on the characteristic ocular and craniofacial findings, a diagnosis of Goldenhar syndrome (Oculo-Auriculo-Vertebral Spectrum) was established.

DISCUSSION

Goldenhar syndrome represents a developmental field defect involving derivatives of the first and second branchial arches.[6] The exact etiology remains uncertain; however, abnormal neural crest cell migration and vascular disruption during embryogenesis have been proposed as possible mechanisms.[7]

The classical triad consists of ocular abnormalities, auricular anomalies, and vertebral defects.[8] Epibulbar dermoid remains the most frequent ocular manifestation and is considered highly characteristic of the syndrome.[9] These lesions are choristomas composed of ectopic dermal tissue containing hair follicles, sebaceous glands, adipose tissue, and connective tissue elements.[10]

Our patient exhibited several classical features including limbal dermoid, facial asymmetry, hemifacial hypoplasia, and preauricular appendage. However, postaxial polydactyly was also present. Limb anomalies are uncommon in Goldenhar syndrome and have been reported only sporadically in the literature.[11] The coexistence of postaxial polydactyly therefore makes the present case noteworthy.

Unlike many reported cases demonstrating vertebral, cardiac, renal, or auditory abnormalities, our patient had no systemic involvement on detailed evaluation. Early diagnosis remains important because affected children may harbor occult congenital anomalies requiring multidisciplinary management.[12]

Management of limbal dermoids depends on lesion size, visual significance, induced astigmatism, and cosmetic concerns. Surgical excision may be considered when visual development is threatened or cosmetic rehabilitation is desired.[13]

CONCLUSION

Goldenhar syndrome should be suspected in any child presenting with limbal dermoid associated with facial asymmetry and preauricular appendages. This case demonstrates a classical ocular and craniofacial presentation with the unusual association of postaxial polydactyly in the absence of systemic abnormalities. Comprehensive multi-disciplinary evaluation remains essential for appropriate diagnosis and management.

Declaration of Patient Consent

Written informed consent was obtained from the patient's parent/legal guardian for publication of clinical details and photographs. Efforts have been made to conceal identity; however, anonymity cannot be guaranteed.

Conflict of Interest

The authors declare no conflict of interest.

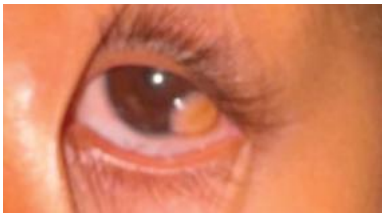

Figure 1. Left Eye Showing Inferotemporal Limbal Dermoid.

Figure 2. Facial Asymmetry with Left Hemifacial Hypoplasia.

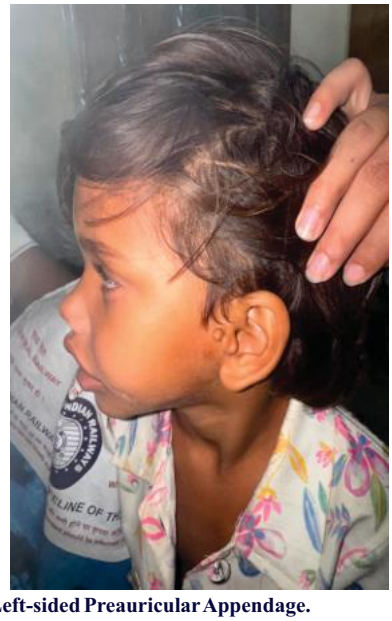

Figure 3. Left-sided Preauricular Appendage.

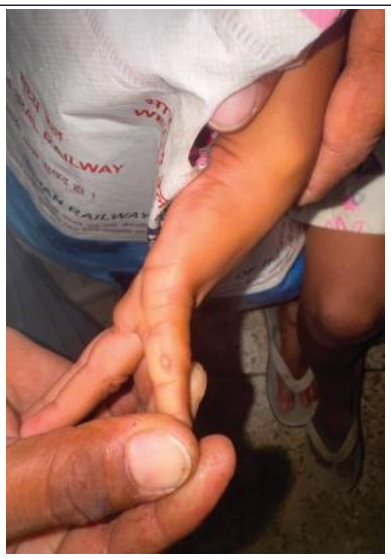

Figure 4. Postaxial Polydactyly of the Hand.

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