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Case Series

Clinicopathological Profile of Limbal Dermoid: A Case Series with Review of Immunohistochemical Findings

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Abstract

Background: Limbal dermoid is a rare, congenital, benign lesion occurring at the corneoscleral junction. Although usually harmless, it can cause cosmetic issues, astigmatism, and occasionally affect vision. Some cases are associated with Goldenhar syndrome, a congenital condition involving craniofacial, auricular, and vertebral anomalies. Histopathology and immunohistochemistry (IHC) help confirm the diagnosis and identify tissue components. This case series describes four Grade I limbal dermoids - two isolated and two linked with Goldenhar syndrome—to highlight their clinical, systemic, and histopathological features.

Methodology: This retrospective case series included four clinically diagnosed limbal dermoid patients evaluated at a tertiary care hospital. All underwent detailed ocular examination, slit-lamp assessment, and photographic documentation. Cases 1 and 4 had isolated limbal dermoids, while Cases 2 and 3 showed features of Goldenhar syndrome. Data collected included demographics, presenting complaints, lesion grade and location, refractive error, systemic findings, and surgical details. All excised specimens were processed and analyzed in our hospital for routine staining and immunohistochemical evaluation.

Discussion: All four patients were 10–13 years old, and all lesions were Grade I, mostly located inferotemporally. Two patients had systemic features of Goldenhar syndrome, such as facial asymmetry and preauricular tags. All children had astigmatism, which improved with refractive correction, and none required urgent surgery. Histopathology showed keratinized epithelium, collagenous stroma, adnexal structures, and adipose tissue in all cases, while syndromic cases also contained cartilage. IHC findings further confirmed the choristomatous nature of the lesions.

Conclusion: This case series highlights the benign nature of Grade I limbal dermoids and their occasional association with Goldenhar syndrome. Most lesions respond well to conservative management, while syndromic cases need thorough systemic evaluation. Histopathology and IHC play an important role in distinguishing isolated lesions from syndromic ones and help guide appropriate treatment planning.

Keywords: Limbal dermoid, Goldenhar syndrome, choristoma, histopathology, IHC, pediatric ophthalmology.

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Introduction

Limbal dermoid is a rare, congenital, and benign growth that develops at the junction between the cornea and the sclera. It is usually detected at birth and slowly increases in size over time. Although non-cancerous, it may cause cosmetic problems and, in some cases, may affect vision if it grows toward the visual axis. Most limbal dermoids occur in only one eye, but in some children, they can be associated with broader systemic abnormalities, including conditions such as Goldenhar syndrome.

Goldenhar syndrome, also called oculo-auriculo-vertebral syndrome (OAVS), is a rare congenital disorder with a wide range of features. It commonly involves eye abnormalities such as limbal dermoids, external ear deformities or preauricular tags, facial asymmetry due to mandibular underdevelopment, and vertebral defects [1]. Dr. Maurice Goldenhar was the first to clearly describe this condition, leading to its recognition as a distinct clinical syndrome [2]. Although uncommon in the general population, it continues to be an important area of interest because of its varied presentation and potential impact on multiple organ systems.

The exact cause of Goldenhar syndrome is not fully understood. It is believed to result from a combination of genetic and environmental factors. Maternal conditions such as diabetes, rubella infection, influenza, heavy alcohol use, and vitamin A toxicity have been linked with the syndrome [3-4]. Exposure to certain drugs during pregnancy—such as thalidomide, retinoic acid, tamoxifen, and even cocaine—has also been reported as a possible risk factor [4-5]. These findings support the idea that both inherited tendencies and external influences during early fetal development may play a role.

Diagnosing Goldenhar syndrome can be challenging because the features vary widely from patient to patient. Many children first come to the Ophthalmology outpatient department with limbal dermoids, making early recognition by eye specialists crucial [6]. Since there are no universally accepted diagnostic criteria, a detailed clinical and systemic evaluation is essential to establish the diagnosis.

Histologically, limbal dermoids contain tissues from ectodermal and mesodermal origins, such as skin, hair follicles, sebaceous glands, and sometimes cartilage. Immunohistochemical (IHC) studies help confirm the diagnosis by identifying the specific tissue components and understanding their patterns of differentiation. These findings provide valuable insights into the biological behavior of the lesion.

In this case series, we present 4 cases out of which two are Goldenhar syndrome and other two are limbal dermoid without any syndrome.

Methodology

Study Design: This study was conducted as a retrospective observational case series comprising four patients diagnosed with limbal dermoid.

Study Setting: The clinical evaluation and documentation of all four cases were performed at our tertiary care hospital. Histopathological and immunohistochemical examinations were performed at our centre, as these facilities are available in the Pathology Department of Narayan Medical College and Hospital, Sasaram. Therefore, all excised specimens were processed and analyzed in hospital for complete histopathological evaluation and immunohistochemical (IHC) analysis.

Case Selection: All four cases included in this study were selected on the basis of a confirmed clinical diagnosis of limbal dermoid, supported by detailed slit-lamp examination and photographic documentation. Case 1 and Case 4 presented with isolated limbal dermoid without any evidence of associated congenital syndromes. In contrast, Case 2 and Case 3 had limbal dermoid in association with Goldenhar syndrome, identified through characteristic craniofacial features and systemic findings consistent with the syndrome. Histopathological and immunohistochemical (IHC) evaluation could be performed at our institution; only those cases with complete clinical records and available histopathology/IHC reports from the external laboratory were included in this case series.

Data Collection: For each patient, comprehensive data were collected, including demographic details such as age and sex, along with clinical information covering presenting complaints, the location and grade of the limbal dermoid, and any associated anomalies. Syndromic evaluation was performed, with detailed assessment for Goldenhar syndrome specifically in Cases 2 and 3. Surgical records, including the type of procedure, operative findings, and any complications, were documented. All excised specimens were preserved in formalin and sent to pathology laboratory, where routine hematoxylin–eosin staining and immune-histochemical (IHC) analysis were performed. The reported IHC markers from the lab were recorded for comparison across the cases.

Case Report

Case 1

Presentation & Clinicopathological Findings: A 10-year-old boy presented with long-standing watering, ocular irritation, and blurring of vision in the right eye. Examination revealed a well-circumscribed Grade I limbal dermoid at the inferotemporal limbus 4.5-5mm approx under slit lamp. Visual acuity was 6/60 unaided, improving to 6/24 with pinhole and BCVA of 6/24; the left eye was normal. Slit-lamp evaluation showed a superficial, creamy-yellow lesion with fine hair follicles.

Investigations & Systemic Evaluation: AS-OCT and ultrasound biomicroscopy confirmed a superficially located dermoid without stromal invasion or involvement of the visual axis.

Comprehensive systemic assessment—including ENT, pediatric, audiometric, skeletal, and dental evaluation—revealed no abnormalities, establishing this as an isolated non-syndromic case. Refractive testing demonstrated correctable regular astigmatism.

Cause of Amblyopia: Primarily due to refractive amblyopia from uncorrected astigmatism. Visual axis not directly obstructed.

Management: The patient was managed by superficial shave excision suitable for grade1 lesions. Topical medications like antibiotic eyedrops 4 to 6 times per day for 7 days. Lubricating eye drops 4 to 6 times per day for 10 days. Mild topical steroid for 7days tapered. The patient was asked to follow up after 1week for epithelial healing. Then 2-4 weeks for refraction and BCVA. And then 3-6months for astigmatism stability, amblyopia response and yearly for recurrence or cosmetic outcome.

Figure of Case 1: limbal dermoid without any syndrome

Case 2

Presentation & Clinicopathological Findings: Case 2 was an 11-year-old girl who presented with a limbal mass in the right eye and decreased vision for several months. Clinical examination revealed a Grade I limbal dermoid measuring about 5-5mm under slit lamp at the inferotemporal limbus. Her visual acuity was 6/60 unaided, improving to 6/24 with pinhole, and a BCVA of 6/18. During general examination, subtle facial asymmetry, preauricular abnormalities, and mild mandibular hypoplasia were noted, raising suspicion of Goldenhar syndrome (oculo-auriculo-vertebral spectrum).

Investigations & Syndromic Features: The patient underwent AS-OCT and USG-BM, both of which showed a superficial lesion without deep stromal involvement. Because of the suspected syndromic features, multidisciplinary systemic evaluation was carried out. ENT assessment, audiometry, dental screening, and vertebral evaluation collectively supported the diagnosis of Goldenhar syndrome, confirming the presence of ocular, auricular, and mild craniofacial anomalies. Refractive error showed a mixture of astigmatism and myopic shift associated with the lesion.

Cause of Amblyopia: Mixed- refractive amblyopia from astigmatism and sensory deprivation was minimal as visual axis not involved.

Management: Simple shave excision was done in right eye followed by topical antibiotic eye drops 4 times per day for 7 days. Lubricating eye drops given 6 times per day for 10days.

Mild topical steroids were given for 7 days by tapering. The patient was asked to follow up after 1 week. Then 2-4 weeks for refraction and BCVA. And then 3-6 months and yearly.

Figure of Case 2: limbal dermoid with Goldenhar syndrome

Case 3

Presentation & Clinicopathological Findings: An 11-year-old girl with a known diagnosis of Goldenhar syndrome presented with a small temporal limbal mass in the right eye and visual blurring. Examination revealed a Grade I limbal dermoid 5-5mm approx. under slit lamp. Visual acuity was 6/24 unaided, improving to a BCVA of 6/18. Slit-lamp evaluation showed a smooth, creamy-white superficial lesion with fine hair-like projections.

Diagnostic Evaluation & Systemic Assessment: AS-OCT and ultrasound biomicroscopy confirmed a superficial dermoid without deep corneal or scleral extension. As part of ongoing syndromic follow-up, updated assessments documented mild preauricular anomalies, minimal conductive hearing changes, and minor vertebral irregularities—findings consistent with oculo-auriculo-vertebral dysplasia. Refractive testing demonstrated low-to-moderate astigmatism attributable to the lesion.

Cause of Amblyopia: Primarily refractive amblyopia, secondary to lesion-induced corneal astigmatism.

Management: The patient was managed by superficial shave excision suitable for grade1 lesions. Topical medications like antibiotic eyedrops 4 to 6 times per day for 7 days. Lubricating eye drops 4 to 6times per day for 10 days. Mild topical steroid for 7days tapered. The patient was asked to follow up after 1week for epithelial healing. Then 2-4 weeks for refraction and BCVA. And then 3-6months for astigmatism stability, amblyopia response and yearly for recurrence or cosmetic outcome.

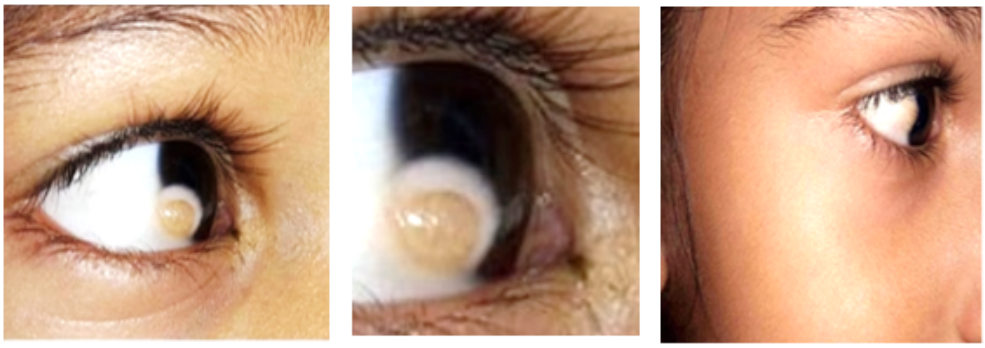

Figure of Case 3: limbal dermoid with Goldenhar syndrome

Case 4

Presentation & Clinicopathological Findings: Case 4 concerned a 13-year-old boy who reported decreased vision in the right and left eye and occasional ocular irritation. Examination revealed a small Grade I limbal dermoid 4.5-5mm approx. under slit lamp, located at the inferotemporal limbus of both eyes. Visual acuity testing showed 6/24 unaided, improving to 6/18 with pinhole, and BCVA was 6/18 both eyes. On slit-lamp examination, the lesion appeared as a superficial, well-circumscribed mass with characteristic dermoid texture.

Investigations & Systemic Screening: AS-OCT and USG-BM imaging confirmed superficial involvement limited to the conjunctival-scleral interface without stromal infiltration. As part of routine evaluation, the patient underwent systemic screening for any syndromic associations. ENT assessment, pediatric evaluation, audiometry, and spinal/dental assessments were all within normal limits, confirming that this was an isolated, non-syndromic limbal dermoid. Refractive evaluation

showed regular astigmatism that partially accounted for her reduced visual acuity.

Cause of Amblyopia: Refractive amblyopia secondary to bilateral astigmatism.

Management: Simple shave excision was done in right eye followed by topical antibiotic eye drops 4 times per day for 7 days. Lubricating eye drops given 6 times per day for 10days.Mild topical steroids were given for 7 days by tapering. The patient was asked to follow up after 1 week. Then 2-4 weeks for refraction and BCVA. And then 3-6 months and yearly.

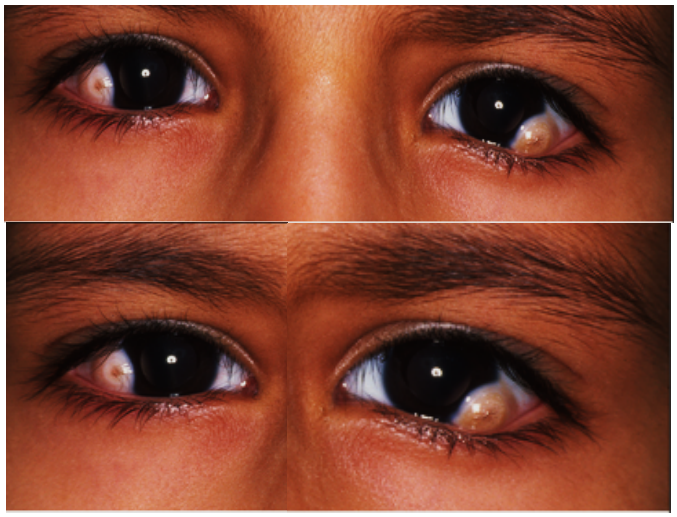

Figure of Case 4: limbal dermoid without any syndrome

Clinicopathological & Immunohistochemical Differences of cases

Clinicopathological Differences

- **Syndromic vs Non-Syndromic:** Cases 2 and 3 were associated with Goldenhar syndrome, showing systemic abnormalities; Cases 1 and 4 were isolated.
- **Lesion Complexity:** All cases had Grade I superficial lesions, but syndromic cases contained cartilage and more complex adnexal tissue, while non-syndromic cases lacked cartilage.
- **Visual and Refractive Findings:** Astigmatism was common; visual acuity varied, with syndromic cases generally showing slightly better baseline vision.
- **Laterality & Symptoms:** Non-syndromic cases included one bilateral lesion (Case 4); syndromic cases had additional cosmetic concerns and facial asymmetry.

Immunohistochemical (IHC) Differences

Non-Syndromic Dermoids (Cases 1 & 4): Standard dermoid profile, showing epidermal and adnexal markers, no cartilage-related markers.

Syndromic Dermoids (Cases 2 & 3): Choristomatous pattern with cartilage differentiation, confirming multi-lineage tissue involvement typical of Goldenhar syndrome.

Table 1: Comparative Summary of Clinical, Systemic, Histopathological Features and Immunohistochemical Findings of the Four Limbal Dermoid Cases

Parameter	Case 1	Case 2	Case 3	Case 4
Age / Sex	10-year-old, Boy	11-year-old, Girl	11-year-old, Girl	13-year-old, Girl
Syndromic Association	None	Goldenhar syndrome	Goldenhar syndrome	None
Eye Involved	Right eye	Right eye	Right eye	Both eye
Lesion Grade	Grade I	Grade I	Grade I	Grade I
Lesion Size	~5–6 mm	~5–6 mm	~2.5–6 mm	<5–6 mm
Presenting Symptoms	Watering, irritation, blurred vision, asthenopia	Blurred vision, ocular lesion, facial asymmetry	Mild blurring of vision, cosmetic concern	Decreased vision, irritation
Unaided Visual Acuity (Affected Eye)	6/60	6/60	6/24	6/24
Pinhole Improvement	Up to 6/9	Up to 6/24	No change	Up to 6/18
Best Corrected Visual	6/24	6/18	6/18	6/9

Acuity				
AS-OCT / USG-BM Findings	Superficial lesion; no stromal invasion	Superficial lesion; no deep invasion	Superficial involvement only	Superficial; no deep extension
Systemic Evaluation	Normal	Features of Goldenhar syndrome	Confirmed Goldenhar features	Normal
Audiometry, ENT, Dental / Vertebral Screening	Normal	Abnormalities consistent with syndrome	Abnormalities consistent with syndrome	Normal
Refractive Error	Astigmatism	Astigmatism + mild myopia	Astigmatism	Astigmatism
Refractive Error (Diopters)	+1.25 D sphere, +1.50 D cylinder @90°	-1.00 D sphere, +2.00 D cylinder @180°	-0.50 D sphere, +1.75 D cylinder @90°	+0.75 D sphere, +1.25 D cylinder @90°
Astigmatism Grade	Grade 1	Grade 2	Grade 1-2	Grade 1
Management	Conservative (lubricants, spectacles)	Conservative: surgical option discussed due to cosmesis	Conservative; surgery optional	Conservative; surgery for cosmetic indication only
Histopathology	Keratinized epithelium, collagen, hair follicles, sebaceous glands, adipose tissue	Keratinized epithelium, adnexal elements + cartilage	Keratinized epithelium, adnexal tissue + cartilage	Keratinized epithelium, collagen, adnexal tissue; no cartilage
IHC Findings	Non-syndromic dermoid profile	Features consistent with choristoma in Goldenhar	Choristomatous pattern with cartilage	Non-syndromic dermoid

Discussion

Limbal dermoids are rare congenital choristomatous lesions arising at the corneoscleral limbus. They are generally benign and slowly progressive, often presenting as cosmetic concerns in childhood. In our series of four cases, all patients were between 10–13 years of age, highlighting the typical early detection of these lesions during school age. Consistent with previous literature, the lesions in all cases were Grade I, confined to the superficial conjunctival-scleral plane, and did not significantly encroach on the visual axis, underscoring the generally benign nature of limbal dermoids [7-8]. The predominance of inferotemporal limbal involvement in our series aligns with previous reports that describe the inferotemporal quadrant as the most frequent site for these lesions [9].

Two of the four cases were associated with Goldenhar syndrome (oculo-auriculo-vertebral spectrum), which agrees with published data suggesting that limbal dermoids may occur as part of a broader systemic congenital disorder. These cases exhibited additional craniofacial anomalies, including facial asymmetry, preauricular tags, mild mandibular hypoplasia, and minor vertebral defects, highlighting the importance of a thorough systemic and multidisciplinary evaluation when a limbal dermoid is detected. Early recognition of syndromic associations is crucial, as management may require input from pediatricians, ENT specialists, orthodontists, and geneticists [10-12]. The non-syndromic cases, on the other hand, had no systemic abnormalities, emphasizing that limbal dermoids may also occur as isolated ocular lesions.

Visual outcomes in this case series were largely favorable, as the lesions were superficial and did not obstruct the visual axis. Refractive errors, particularly astigmatism, were noted in all patients and were likely induced by the corneal distortion caused by the lesion. Visual acuity improved with correction in most cases, indicating that early refractive assessment is essential to prevent amblyopia or school-related difficulties in children[13]. None of the patients had significant visual impairment requiring urgent surgical intervention, supporting the concept that conservative management is appropriate for small, non-progressive lesions, with surgery reserved for cosmetic reasons, progressive astigmatism, or potential amblyopia.

Histopathological findings in all cases confirmed the typical composition of limbal dermoids, including keratinized stratified squamous epithelium, dense collagenous stroma, and adnexal structures such as hair follicles, sebaceous glands,

and adipose tissue. Notably, the Goldenhar-associated cases showed the presence of mature cartilage within the dermoid, a characteristic feature of syndromic limbal dermoids. Immunohistochemical evaluation further supported these observations, demonstrating differential tissue expression patterns consistent with choristomatous lesions. These findings corroborate the existing literature indicating that limbal dermoids represent a developmental anomaly of ectodermal and mesodermal origin, with choristomatous elements sometimes extending to cartilage in syndromic cases[7, 10].

Compared with published literature, our series supports the predominance and relatively benign nature of Grade I limbal dermoids, as reported by Zhong et al. 2018 [14], who found that 59 % of dermoids were classified as Grade I with better visual prognosis than higher grades, reflecting the superficial lesions and favorable outcomes seen in our patients. Al Ghadeer et al. 2023 [15] documented that astigmatism greater than 1 D was present in a majority of limbal dermoid eyes, emphasizing the impact of corneal distortion on refractive error and amblyopia, which parallels the astigmatic refractive errors and amblyopia observed across our cases.

Our study further adds value by incorporating immunohistochemical (IHC) analysis, with markers such as Ki67, S-100, and SOX9, which were crucial in differentiating cartilage-containing syndromic dermoids from non-syndromic lesions, even when systemic features were subtle or absent. This approach allowed early recognition of syndromic cases, guided appropriate systemic evaluation, and facilitated timely surgical planning. By correlating clinical features, refractive errors, amblyopia, and histopathological findings with IHC results, our study provides a more comprehensive understanding of both syndromic and non-syndromic Grade I limbal dermoids, supporting individualized management and optimal visual and cosmetic outcomes.

From a management perspective, the decision-making process in our series emphasized individualized care. All four patients were initially managed conservatively with lubricating eye drops and refractive correction. Surgical intervention, when considered, focused on preserving limbal stem cells and ocular surface integrity, with the potential use of amniotic membrane grafting in cases where excision might involve deeper tissues.

This approach aligns with current recommendations in pediatric ophthalmology, which advocate for a careful balance between functional preservation and cosmetic improvement [8].Overall, this case series highlights the spectrum of clinical, systemic, and histopathological features of Grade I limbal dermoids. It underscores the importance of comprehensive evaluation, particularly for early detection of syndromic associations, and demonstrates that most lesions can be managed conservatively with satisfactory visual and cosmetic outcomes. The presence of cartilage in Goldenhar-associated dermoids emphasizes the value of histopathological and immunohistochemical assessment in differentiating isolated from syndromic lesions, which can guide counseling, systemic evaluation, and surgical planning.

Conclusion

This case series highlights the clinical spectrum of Grade I limbal dermoids, demonstrating their predominantly benign and superficial nature, with two cases occurring in isolation and two associated with Goldenhar syndrome. Although all four lesions shared similar ocular characteristics, the syndromic cases showed additional systemic anomalies requiring multidisciplinary evaluation. Immunohistochemical (IHC) analysis, particularly using markers such as Ki67, S 100, and SOX9, proved invaluable in differentiating syndromic from non-syndromic lesions, even when systemic features were subtle or not yet clinically apparent.

Conservative management was sufficient in most patients, with surgical intervention reserved for cosmetic, symptomatic, or refractive indications. Histopathology across all specimens confirmed the typical choristomatous components of limbal dermoids, while the Goldenhar-associated cases showed additional cartilaginous elements consistent with their syndromic background.

Overall, early IHC evaluation, appropriate systemic screening, and individualized management are essential for timely diagnosis, optimal visual rehabilitation, and improved cosmetic outcomes in children with limbal dermoid.

Conflict of Interest Statement: The authors declare that there is no conflict of interest related to this study.

Informed Consent and Human Rights Statement: Informed consent was obtained from the parents or legal guardians of all patients included in this case series. All procedures performed were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments.

Authorization for the Use of Human Subjects / Ethical Approval: The study protocol involving human subjects was reviewed and approved by the Institutional Ethics Committee of our tertiary care hospital. The research adhered to all relevant

national regulations and institutional policies concerning human subjects.

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