Original Research Paper

Volume - 14 | Issue - 03 | March - 2024 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Ophthalmology

BILATERAL GOLDENHAR SYNDROME: A RARE CASE REPORT

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ABSTRACT **Objective:** To highlight bilateral characteristics in a young female with a rare incidence of Goldenhar syndrome. **Method:** The development of the eye, ear, face, and vertebral column is impacted by the unusual congenital condition known as Goldenhar syndrome. One in every 3,500–5,600 births are impacted by it. The abnormality often only affects one side of the body. A 19-year-old female presented with a major complaint of a mass in her right eye and visual loss in both eyes since birth. There was a history of cleft lip, cleft palate, wide nasal bridge, preauricular tag on the left side, absent right ear (anotia), and cleft lip and palate. Vertebral and cardiac abnormalities have never existed. On inspection, there were microcornea, limbal dermoid, and conjunctival dermoid in the right eye, and visual acuity of 6/60 in the right eye and 6/36 in the left eye. Fundus examination reveals a small disc in the right eye whereas grade 4 fundal coloboma in the left eye does not involve the disc and macula. ENT examination reveals an atretic external auditory canal and absent tympanic membrane but the presence of tragus on the right side whereas a pre-auricular tag in the left ear. B-scan shows findings were consistent with fundal coloboma in the left eye. Chest x-ray and 2D-ECHO were normal. **Result:** With best-corrected visual acuity, refractive error correction is provided. 6/36 for the right eye and 6/6 for the left. Artificial tear drops to alleviate dryness. Craniofacial surgery was performed since the patient had significant cleft lip, cleft palate, and macrognathia. **Conclusion:** Goldenhar Syndrome is a rare congenital condition characterized by cosmetic problems that require a comprehensive approach for successful care. In most cases, it is unilateral, but it can be bilateral.

KEYWORDS : Goldenhar syndrome, limbal dermoid, anotia, cleft palate.

INTRODUCTION

Goldenhar syndrome is a rare congenital disorder that affects the development of the eyes, skull, head, face, and vertebral column. Also known as oculo-auriculo-vertebral dysplasia. Goldenhar syndrome affects one out of every 3,500 to 5,600 children at their time of birth with a male-to-female ratio of 3:2.

This illness can occasionally impact internal organs as well. The degree of Goldenhar syndrome varies from instance to case. The ailment falls under the category of illnesses known as craniofacial microsomia. However, some study suggests that it results from problems with blood circulation or limitations on embryonic growth. Additionally, research suggests that genetics may be involved since a tiny proportion of individuals with Goldenhar syndrome may have relatives who also exhibit the disorder. It is usually sporadic.

Ocular features, apart from an epibulbar dermoid, include upper lid notching or coloboma, microphthalmos, and fundal or disc coloboma. Systemic features include hypoplasia of the malar, maxillary, and mandibular regions, macrostomia and microtia, preauricular and facial skin tags, hemivertebrae (usually cervical), mentally disabled, cardiac, renal, and central nervous system (CNS) anomalies.

Goldenhar syndrome often only impacts one side of the face. But occasionally, it might have an impact on both sides. Aside from these symptoms, people with Goldenhar syndrome may also experience hearing loss, lung issues, limb deformities, spinal issues, and kidney issues.

MATERIALS AND METHODS:

Case Details

A 19-year-old female presented with a complaint of limbal mass with hair follicles in her right eye since birth. Associated with diminution of vision in both eyes. There was a history of cleft palate and cleft lip reconstruction 15 years back. With no family history of maternal infections or illness during pregnancy. No history of similar complaints among family members.

She was well-built and well-nourished with no co-morbidities and vitals are within normal limits. Her general examination reveals Bilateral Cleft lip, Cleft palate, Anotia (absent right pinna) with the presence of tragus, a Preauricular skin tag in the left ear, Broad nasal bridge, and Macrostomia.

On ocular examination, visual acuity in the right eye is 6/60, and the left eye is 6/18. Right eye shows epibulbar limbal mass of size 2x2 cm present temporally over conjunctiva extending up to limbus, soft in consistency with ill-defined margin, elliptical, mobile, non-pulsatile, translucent, pink in color, presence of hairs over surface 2x2 cm over an inferotemporal quadrant of the conjunctiva. The surface is irregular, and microcornea and black hairs are present.

Another swelling of 1.5x1x1 cm soft, cystic reddish blue in color present over palpebral conjunctiva over lateral 1/3 of upper lid with ill-defined margin and surface is irregular. Non-mobile, non-tender, non-pulsatile translucent mass. Microcornea, rest within normal limits.

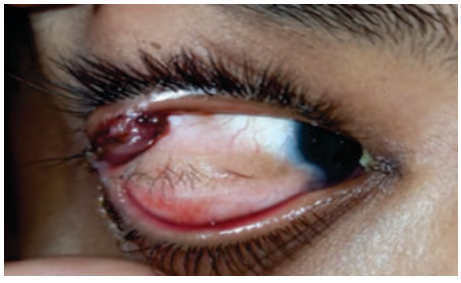

Figure 1: Epibulbar limbal dermoid with hairs in right eye

Investigation

All routine blood investigations were normal. Figure 3: Fundus photograph showing fundal coloboma in left eye. Fundus examination of left eye reveals Grade 4 fundal coloboma of size approx. 2-3 DD, circular in shape, 1 Disc Diameter away from disc inferonasal in left eye sparing disc and macula. The B-scan of the right eye is within normal limits whereas the left eye shows fundal coloboma inferiorly. Chest X-ray, Echocardiogram, and CT scan show no abnormality.

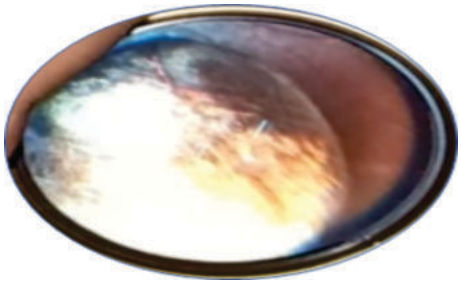

Figure 3: Fundus photograph showing fundal coloboma in left eye.

RESULTS

Refractive error correction is given: Spectacles with best corrected visual acuity 6/36 in the right eye and 6/6 in the left eye with regular follow-up. Rehabilitation strategies like Hearing aids for hearing loss and surgical reconstruction of the pinna. For Bilateral Cleft palate and cleft lip: surgical reconstruction was done.

DISCUSSION

The abnormalities are usually unilateral in 85% of cases and bilateral in 10-33% of cases. In Goldenhar syndrome, ocular anomalies, especially bilateral dermoids seen in 60% of cases, vertebral anomalies (40%), and ear anomalies (40%) cases.

The male-to-female ratio is 3:2, with the right side being more frequently affected than the left with a ratio of 3:2. Tiple et al. 2015 found bilateral features in a seven-year-old patient like accessory tragi, bilateral ocular dermoid, and mandibular hypoplasia (micrognathia). M. Bedi et al. found right macrostomia, bilateral preauricular skin tags, bilateral CTEV, squint in bilateral eyes, thoracic vertebral anomalies, right-sided aortic-arch, and associated left pulmonary agenesis in an 18-month-old male child. Similarly, P. Bajaj et al.in 2017 found in a seven-year-old male bilateral epibulbar dermoid, mild conductive deafness, preauricular tags, and operated cleft lip.

CONCLUSION

Goldenhar Syndrome is a rare congenital anomaly, with cosmetic defects whose treatment may pose numerous challenges with a multidisciplinary approach for optimal management. It usually affects unilaterally in most cases but it can be bilateral. The timing of the reconstruction plays a crucial role in the treatment. Primary reconstruction typically consists of a cleft repair, corrections of colobomas, ear deformities, and dermoid and preauricular tags removal. The comprehensive treatment focuses not only on dental care, articulation, and hearing but also on the prevention and treatment of the malformation's psychosocial elements. Treatment necessitates ongoing monitoring and re-evaluation of outcomes.

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