

Case Report: Goldenhar Syndrome with Atypical Features

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Abstract

Significance: Amblyopia is typically overlooked in patients with Goldenhar Syndrome (GS) but identifying and correcting the amblyogenic factors may result in a better visual outcome. We present a case of GS with unique ocular findings of pigment dispersion, horizontal nystagmus and preretinal bands.

Purpose: To report a case of Goldenhar syndrome with unique ocular findings.

Case Report: An eight-year-old female Asian patient with complaint of decreased vision for four years reported to our office. Examination revealed compound myopic astigmatism with inter-ocular disparity, eyelid, iris and retinochoroidal colobomata along with preauricular facial tags, the features characteristic of Goldenhar syndrome. However, the findings of pigment dispersion, attached high frenulum, horizontal jerk nystagmus and pre-retinal bands were unique to the reported case. With spectacle correction and amblyopia therapy, the visual acuity gradually improved from 4/60 and 3/60 to 6/18 and 6/24.

Conclusion: Goldenhar syndrome has a multitude of associated abnormalities. Our case was unique owing to the presence of various uncommon features. Furthermore, individuals with GS can develop amblyopia and the role of eye care professionals (Optometrists and Ophthalmologists) is to provide correction of refractive errors, amblyopia management and to manage associated abnormalities of ocular structures.

Keywords: Goldenhar Syndrome; Oculo-Auriculo-Vertebral Spectrum; Coloboma; Amblyopia

Introduction

Goldenhar (1952) described a syndrome with epibulbar dermoid, preauricular appendages and pretegral fistula. It is a spectrum of dysmorphogenesis of first and second brachial arches. Also known as Oculo-auriculo-vertebral spectrum; a rare syndrome affecting 1 out of 5600 to more than 20,000 live births with a predilection for males by 3:2 [1].

The etiology and pathogenesis of GS is subject to controversy among authors. It clearly reflects etiological heterogeneity due to variable presentation and expression. Both autosomal dominant and autosomal recessive patterns of inheritance have been reported however, most of the cases are sporadic [2].

The diagnosis of Goldenhar syndrome is mainly based on clinical grounds and characterized by presence of ocular, auricular and vertebral anomalies (oculo-auriculo-vertebral spectrum). Clinical features include maxillary, mandible and malar hypoplasia, microtia, periauricular tags, hemivertebrae, mental deficiency, microphthalmos, epibulbar dermoid and lid and disc coloboma.

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Report of Case

An eight-year-old girl was brought visited our office with a chief complaint of decreased vision for four years. The parent also had noticed an abnormality of right upper lid shortly after birth. The patient was previously seen by three eye care doctors (two ophthalmologists and one optometrist). None of the previous doctors had offered any definitive diagnosis however, the parents were told by each doctor that the problem couldn't be corrected due to some defect of posterior part of eyes. Her birth, medical, family medical and family ocular was insignificant.

Her entering visual acuity was 4/60 O.D and 3/60 O.S which were corrected to 5/60 O.U with best correction -5.00DS/ -2.00 DC X 180 OD and -7.00DS/ -2.50DC X 180 OS. Confrontational visual fields to careful finger counting showed superior constriction.

No facial asymmetry was noted. She had multiple preauricular tags, normal external and internal ears, macrostomia and an attached high frenulum.

Ocular examination revealed telecanthus, full unrestricted extraocular motilities, and bilateral fine horizontal jerk nystagmus. Furthermore, a coloboma at medial 1/4th of right upper lid was noted. Slit lamp examination revealed microcornea (7 mm horizontal x 9 mm vertical) OU, iris coloboma at 6 clock position with keyhole configuration OU, subtle pigment dispersion in anterior chamber OS. Scleral Spur was most posterior structure identifiable on gonioscopic examination, along with a hyperpigmented Trabecular Meshwork circumferentially. On dilated fundus examination, there were vitreous liquefaction and bands OU, large inferior retinochoroidal colobomas with partial involvement of maculae OU, indistinct optic disc margins with overlying glial tissue OU, and preretinal fibrous bands OD.

Detailed review of systems and internal ear examination were unremarkable.

The patient was diagnosed with Goldenhar syndrome. She was corrected for her refractive error and advised for a continuous spectacle wear. Three months later, her VA had improved to 6/18 OD and 6/36 OS. A full time, 4 hours daily patching of right eye was advised.

After another three months, the recorded BCVAs were 6/18 OD, 6/24 OS. Patient didn't improve beyond 6/18, 6/24; and patching was discontinued, 3 months after the best possible visual acuity was gained. Her IOP measurement over the entire period remained stable around 12 to 14 mmHg OU. Any variation in magnitude of pigment dispersion was not observed over the entire period of follow up.

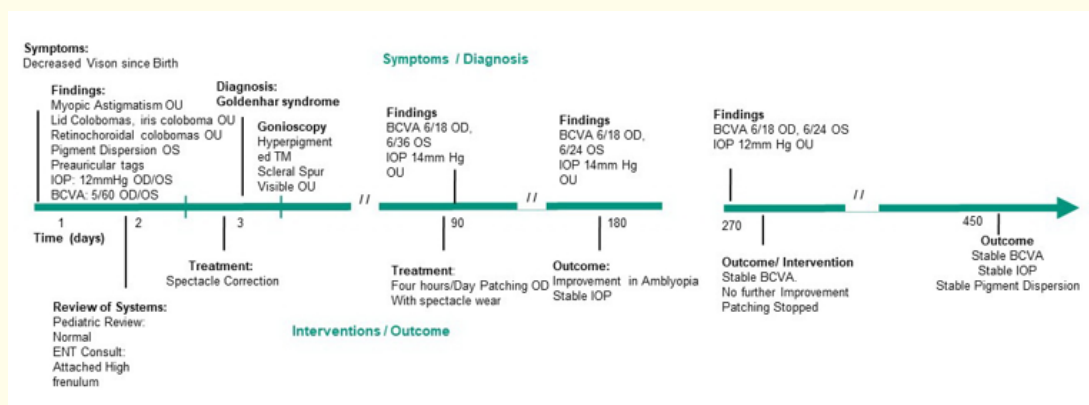


Figure 1: Clinical Timeline: 8-year old Asian female diagnosed with Goldenhar syndrome. IOP= intraocular pressure; BCVA= best corrected visual acuity; TM= trabecular meshwork = idiopathic intracranial hypertension.



Figure 2: A) Attached frenulum, B) preauricular tags, C) Telecanthus.

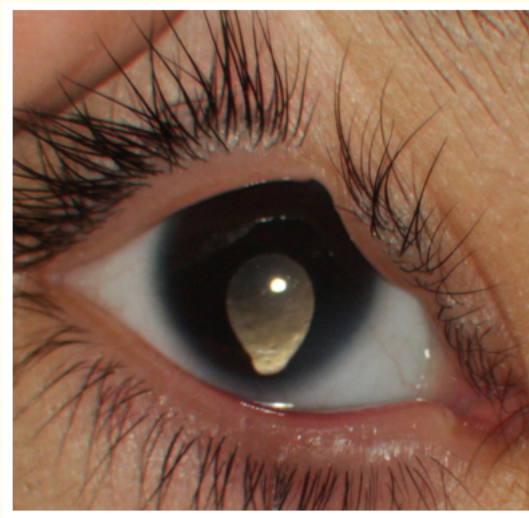


Figure 3: Coloboma of upper lid and Inferior Iris OD.

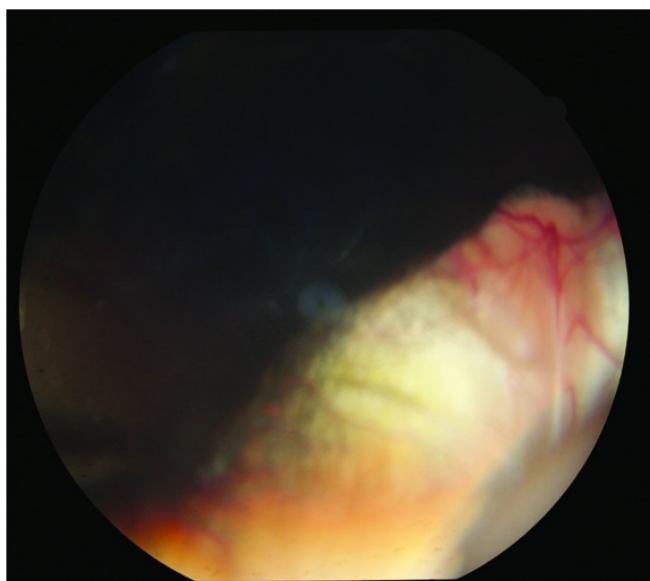


Figure 4: Right large inferior retinochoroidal coloboma and preretinal band along inferior aspect of optic disc

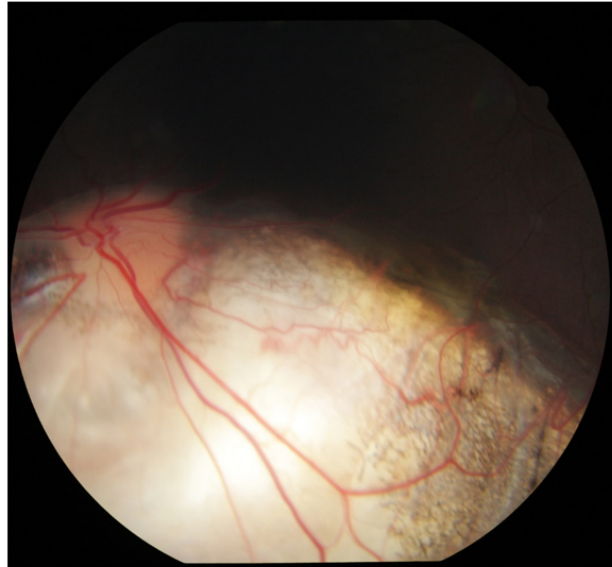


Figure 5: Left large inferior retinochoroidal coloboma.

Discussion and Conclusion

Goldenhar syndrome has a wide spectrum of associated anomalies. No minimal diagnostic criterion has been defined and agreed. However few authors have proposed classification and disease severity grading [3]. Ocular features of Goldenhar syndrome include anophthalmia, micro-ophthalmia, coloboma of upper lid, iris and choroid, epibulbar dermoid, tilted disc, optic nerve hypoplasia, heterotopia of macula and retinal vascular tortuosity [4].

In a study of 18 patients with Goldenhar syndrome, Strömmland., *et al.* [5] observed ear abnormalities in 100% of patients, ocular abnormalities in 72%, vertebral malformations in 67%, cerebral anomalies in 50% and congenital heart defects were found in 33% of patients. In same study he observed hearing impairment (83%), visual deficit (28%), feeding/eating difficulties (50%), speech difficulties (53%), mental retardation (39%) and severe autistic features in 11% of study patients. A multidisciplinary approach is necessary to detect any undiagnosed anomaly of systems. We mandate a complete review of systems by an Optometrist, Otolaryngologists and a Pediatrician.

Ophthalmic findings atypical in our case are nystagmus, pigment dispersion and pre-retinal bands. Although downbeat nystagmus as sign of chiari malformation has been reported to be a rare association of goldenhar syndrome, but horizontal jerk nystagmus has never been reported. In this case it might occurred due to poor vision of patient and involvement of macula and Optic discs by coloboma [6].

None of the authors have reported pigment dispersion in patients with goldenhar syndrome. In the reported case, it may be an unusual association or an unrelated insult. It might occur because as in this patient with microcorneae there were open angles. The posteriorly displaced irides would rub the lens causing pigment dispersion in anterior chamber [7].

Preretinal bands have never been reported in literature which also may be a rare manifestation of goldenhar syndrome or an isolated finding in this patient.

The patients with Goldenhar syndrome are also subject to amblyogenic factors i.e. uncorrected refractive errors along with other ocular and systemic co-morbidities. This patient responded well to the amblyopia therapy.

Refractive errors explained in goldenhar syndrome are usually astigmatic errors due to epibulbar dermoids disturbing normal corneal surface. The refractive errors, particularly anisometropia and astigmatism are well known to cause amblyopia and same mechanism was responsible for development of amblyopia in our patients. In cases with remarkable ophthalmic pathologies and ocular developmental disorders correction of refractive errors and amblyogenic factors are mostly overlooked. Identifying coexisting amblyogenic factors and treating them may result in a better visual outcome.

Patients with Goldenhar syndrome should undergo complete ocular and systemic workup. The role of eye doctors (Optometrists and Ophthalmologists) is to monitor for manage any epibulbar dermoid, correction of refractive error, amblyopia therapy and other potential complications that may develop over time. They are also responsible to make appropriate referrals to related specialties if initial diagnosis or goldenhar syndrome suspected by them.

Disclosure

The authors have no financial or proprietary interests to disclose.

Informed Consent

Written informed consent was obtained from the parents for use of potentially identifiable content in scholarly work.

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