Anterior segment dysgenesis and secondary glaucoma in Goldenhar syndrome

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A 3-year-old girl presented with a history of watering, haze and increase in the size of the right eye for two months. The child had bilateral preauricular skin tags, limbal dermoid and dermolipoma, consistant with the diagnosis of Goldenhad syndrome. In addition, her right eye manifested enlarged cornea, flat anterior chamber, atrophic iris and elevated intraocular pressure. This case report highlights a possible association of anterior segment dysgenesis and glaucoma with Goldenhar syndrome.

Key words: Anterior segment dysgenesis, dermolipoma, Goldenhar syndrome, limbal dermoid, preauricular skin tags, secondary glaucoma

Case Report

Anterior segment dysgenesis is a spectrum of disorders which includes maldevelopment of anterior segment structures including the cornea, anterior chamber angle, and iris due to defective neural crest cells migration or differentiation. Goldenhar syndrome is a congenital craniofacial syndrome, and the classical triad includes epibulbar dermoid, preauricular skin tags, and vertebral anomalies. An association of the two has not been reported. Presented here is a patient with classical features of Goldenhar syndrome (bilateral preauricular skin tags, bilateral limbal dermoids, and dermolipoma) along with anterior segment dysgenesis and glaucoma in one eye.

A 3-year-old girl was brought by her parents with complaints of watering, corneal haziness, and an increase in the size of the right eye noticed for the last two months. The child was born by a non-consanguineous marriage with normal vaginal delivery. There was no history of antenatal exposure to drugs or gestational diabetes mellitus. There was no history of previous complaints/ocular trauma and no abnormality was noted in the eye in past.

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The child had congenital preauricular skin tags on both sides [Fig. 1]. Torchlight examination revealed an enlarged hazy cornea in the right eye [Fig. 2a] and a limbal dermoid in inferotemporal limbus of left eye [Fig. 2b]. The visual acuity in the right eye was hand movement close to face with inaccurate projection of rays and 6/12 in the left eye on Snellen chart. Refraction under cycloplegia for the left eye was +0.5DS/-0.5DC × 140° which was non-significant, and the patient was not given any refractive correction.

On slit lamp evaluation, the right eye had a flat anterior chamber with atrophic iris and absence of a pupil with no view of the posterior segment. The intraocular pressure (IOP) was digitally high, and antiglaucoma eye drops (Brinzolamide1% tds and Timolol 0.5% bd) were started. The left eye had normal anterior and posterior segments with healthy optic disc with cup-disc ratio of 0.4. Ultrasonography of right eye revealed an echo-free vitreous cavity with attached retina and optic nerve head cupping.

On Examination under anesthesia (EUA), IOP was 30 mm and 14 mm Hg by Perkins tonometer (Haag-Streit, Koeniz, Switzerland) in the right and left eye respectively. The right eye [Fig. 3a] had an inferotemporal limbal dermoid, a dermolipoma along with hair follicles in the superotemporal conjunctiva extending up to the fornix and intercalary staphyloma superiorly. The anterior chamber (AC) was flat; the iris was atrophic with no discernible pupillary opening. The left eye had two inferotemporal limbal dermoids and a dermolipoma with hair follicles in the temporal bulbar conjunctiva extending up to the fornix [Fig. 3b]. The rest of the eye examination was normal.

The corneal diameter was 15 mm and 12.5 mm, and Axial length on A-scan ultrasonography was 24.4 and 20.2 mm in the right and left eye, respectively. Ultrasound biomicroscopy (UBM) of the right eye revealed iridocorneal as well as iridolenticular adhesions [Fig. 4].

The presence of bilateral preauricular skin tags since birth, bilateral limbal dermoids, and dermolipoma was consistent with the diagnosis of Goldenhar syndrome. On systemic evaluation, there were no abnormalities in the vertebral, respiratory, cardiovascular, or genitourinary systems. X-ray thoracolumbar spine was advised after paediatric consultation to rule out vertebral anomalies which was not done by the parents because of poor follow-up. The presence of severe anterior segment dysgenesis with iridocorneal adhesions, intercalary staphyloma, optic nerve head cupping on ultrasonography, and low vision, resulted in a decision against incisional surgery in the right eye. A 270-degree diode laser cyclophotocoagulation was done under general anesthesia. The left eye was kept under observation. On Follow-up EUA 3 months later, the IOP was 22 mm Hg and 16 mm Hg in the right and left eyes respectively without any

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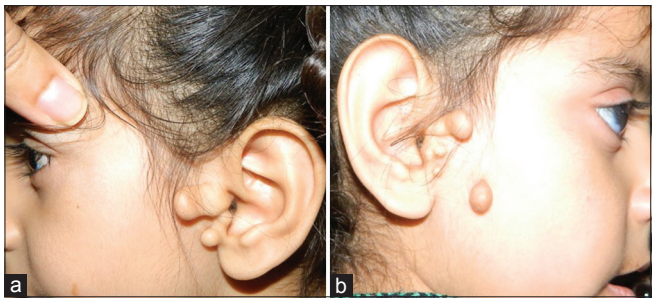

Figure 1: (a) and (b) Preauricular skin tags on both sides of face

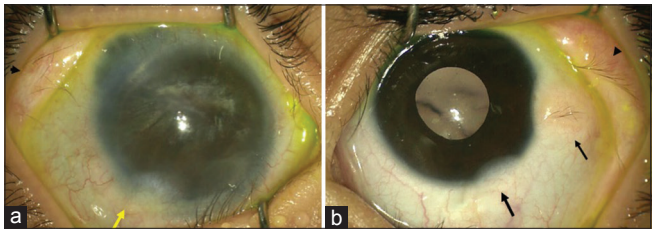

Figure 3: (a) Right eye photograph showing limbal dermoid in inferotemporal limbus (arrow), dermolipoma in superior conjunctiva extending upto the fornix (arrowhead) and intercalary staphyloma superiorly. (b) Left eye photograph showing two limbal dermoids in inferotemporal limbus (arrows), dermolipoma in superior bulbar conjunctiva extending upto the fornix (arrowhead)

antiglaucoma medications. The child is under observation with controlled IOP on one topical antiglaucoma medication.

Discussion

Goldenhar syndrome, also known as the oculo-auriculo-vertebral disease, is a congenital disease with a classic triad of auricular, ocular, and vertebral anomalies. It occurs due to defect in the first and second branchial arch and is mainly sporadic. Genetic inheritance has been described in 1–2% of the cases with the autosomal dominant or recessive pattern.[1] The exact etiology of the disease is not known. The various hypotheses are exposure to drugs/chemicals during pregnancy, gestational diabetes mellitus, chromosomal abnormalities, vascular disturbance, or neural crest cell disorder.[2-4] Different chromosomal anomalies have been reported, including trisomy 7, trisomy 22, Turner's syndrome, etc.[1,5-7] A variety of systemic and ocular findings have been described in this syndrome, the pathogenesis for which is still not clear.

Systemic features reported with Goldenhar's syndrome include mandibular hypoplasia, cleft lip, cleft palate, macrostomia, micrognathia, short neck, neck webbing, cardiovascular abnormalities (in the form of ventricular septal defects, tetralogy of Fallot), urological abnormalities, umbilical hernia, undescended testis, vaginal abnormalities, etc.[8]

Apart from limbal dermoids, many other ophthalmic features have also been reported including colobomas involving eyelids, iris and/or fundus, optic nerve hypoplasia, macular hypoplasia, tilted optic discs, microphthalmos, etc.[9] There is one report which mentions the association of Goldenhar syndrome with juvenile open-angle glaucoma in a patient with Turner's syndrome.[7]

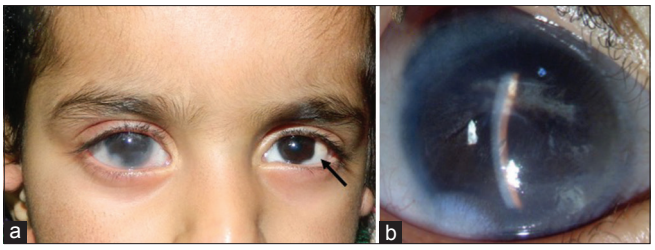

Figure 2: (a) Facial photograph showing enlarged and hazy cornea of the right eye and clear cornea with limbal dermoid at inferotemporal limbus of the left eye (arrow). (b) Slit-lamp photograph of the right eye showing flat anterior chamber with atrophic iris and absence of pupillary opening

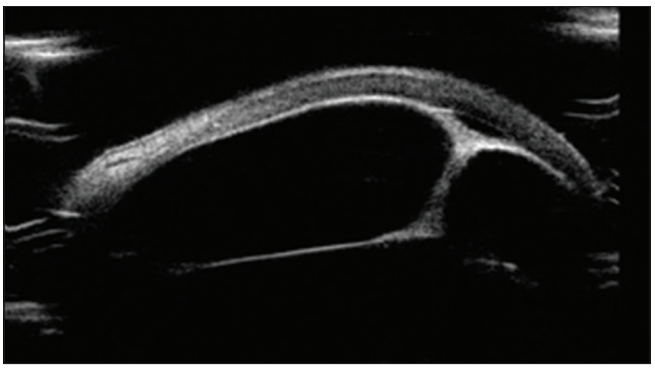

Figure 4: Ultrasound biomicroscopy showing flat anterior chamber with iridocorneal and iridolenticular adhesions

There has been no previous report of anterior segment dysgenesis with glaucoma in Goldenhar syndrome. Karyotyping revealed 46 XX which ruled out Turner's syndrome. We based our diagnosis on clinical grounds as further genetic analysis and whole genome sequencing was not done as parents were not willing and lost to follow-up subsequently. Since neural crest cells are very important for the formation of the anterior segment of the eye,[10] the hypothesis of a neural crest cell disorder seems plausible for our patient. Although genetic disorders can have a variety of ocular and systemic findings, it is also possible that they are just co-incidental. ASD could have incidentally occurred as separate entity from the Goldenhar syndrome in our patient.

As the exact etiopathogenesis of Goldenhar syndrome is still uncertain, each associated finding with the disease deserves to be reported so as to aid in exploring the disease pathogenesis and enhance our understanding of the evolution of the disease. For each case of Goldenhar syndrome, a detailed ocular, and systemic evaluation is very important to detect and treat the associated disease at the earliest possible time. Anterior segment dysgenesis with glaucoma can possibly be one of the ocular findings in a patient with Goldenhar syndrome apart from the other findings of limbal dermoid, dermolipoma, microphthalmos, etc.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other

clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

References

1. Pridjian G, Gill WL, Shapira E. Goldenhar sequence and mosaic trisomy 22. Am J Med Genet 1995;59:411-3.
2. Boles DJ, Bodurtha J, Nance WE. Goldenhar complex in discordant monozygotic twins: A case report and review of the literature. Am J Med Gen 1987;28:103-9.
3. Araneta MR, Moore CI, Onley RS, Edmonds LD, Karcher JA, McDonough C, *et al*. Goldenhar syndrome among infants born in a military hospital to Gulf war veterans. Teratology 1997;56:244-51.
4. Snider TN, Mishina Y. Cranial neural crest cell contribution to the craniofacial formation, pathology, and future directions in tissue engineering. Birth Defects Res C Embryo Today 2014;102:324-32.
5. Stanojevic M, Stipoljev F, Koprcina B, Kurjak A. Oculo-auriculovertebra (Goldenhar) spectrum associated with pericentric inversion 9: Coincidental findings or etiological factor? J Craniofac Genet Dev Biol 2000;20:150-4.
6. Wilson GN, Barr M Jr. Trisomy 9 mosaicism: Another etiology for the manifestations of Goldenhar's syndrome. J Craniofac Genet Dev Biol 1983;3:313-6.
7. Rao VA, Kaliaperumal S, Subramanian T, Rao KR, Bhargavan R. Goldenhar's sequence with associated juvenile glaucoma in Turner's syndrome. Indian J Ophthalmol 2005;53:267-8.
8. Ashokan CS, Sreenivasan A, Saraswathy GK. Goldenhar syndrome - review with case series. J Clin Diagn Res 2014;8:ZD17-9.
9. Margolis S, Aleksic S, Charles N, McCarthy J, Greco A, Bdzilovich G. Retinal and optic nerve findings in Goldenhar-Gorlin syndrome. Ophthalmology 1984;91:1327-33.
10. Sowden JC. Molecular and developmental mechanisms of anterior segment dysgenesis. Eye 2007;21:1310-8.