Management of a rare case of Goldenhar syndrome

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Abstract

We report here a case with the classic signs of Goldenhar syndrome in the form of multiple accessory tragi, bilateral ocular dermoids, lid coloboma, pre-auricular tragi.

Keywords: Accessory tragus, Epibulbar dermoids, Goldenhar syndrome.

Introduction

Goldenhar syndrome (oculoauriculovertebral dysplasia with hemi facial microsomia) is a rare congenital developmental anomaly involving the first and second branchial arches. The classic features of this syndrome include ocular changes such as microphthalmia, epibulbar dermoids, lipodermoids and coloboma; aural features such as preauricular tragi, hearing loss and microtia; and vertebral anomalies such as scoliosis, hemi vertebrae and cervical fusion. In addition to facial, vertebral, ophthalmic malformations cardiovascular, CNS, genitourinary malformations have been associated with goldenhar syndrome. It is a disorder where the patient's facial features are incompletely developed on one side, resulting in eye, ear, and jaw abnormalities. In 85% of patients with Goldenhar syndrome, only one side of the face is affected. Cervical spine vertebral deformities are part of the collection of symptoms. The syndrome was first described in 1952 by the French ophthalmologist Maurice Goldenhar[1] the incidence of Goldenhar syndrome has been reported to be between 1:3500 and 1:5600, with a male: female ratio of 3:2.[2] The exact etiology is not known. However, it is possible that abnormal embryonic vascular supply, disrupted mesodermal migration or some 0other factor leads to defective formation of the branchial and vertebral systems.[2,3] Most of the cases have been sporadic. Autosomal dominant, autosomal recessive, and multifactorial modes of inheritance have also been suggested.[2]

The classic features of this syndrome include ocular changes such as microphthalmia, epibulbar dermoids, lipodermoids, and coloboma; aural features such as pre-auricular tragi, hearing loss, and microtia; and vertebral anomalies such as scoliosis, hemivertebrae, and cervical fusion.[1,4] The abnormalities are found to be unilateral in 85% of cases and bilateral in 10-33% cases.[4] In Goldenhar syndrome, ocular anomalies especially bilateral dermoids are seen in 60% of the cases, vertebral anomalies in 40% of the cases, and ear anomalies also in 40% of the cases.[2] Other systemic features are found in about 50% of the patients.[5] Tetralogy of Fallot and ventricular septal defects are the most common cardiovascular anomalies associated with OAVS[4]. Cleft lip and palate, macrostomia, micrognathia, webbing of the neck, short neck, Tracheoesophageal fistula, abnormalities of sternocleidomastoid muscle, umbilical hernia, inguinal hernia, urologic anomalies, hypoplastic vagina, and anal anomalies may be associated.[4]

Case Report

Eight month old male child born of non-consanguinous parents was brought with abnormalities of the eyes and ears since birth. The child was delivered normally at full term. The antenatal, intranatal and postnatal periods were uneventful. All other family members were normal. On examination, the child was markedly undernourished weighed only 1.9 kg and showed the following abnormal feature. The following ocular features were present: Right eye showed coloboma in the upper eyelid, with two epibulbar dermoids. the temporal dermoid was covering more than half of the cornea and extending a few mm on to the sclera and the nasal dermoid was covering one third of the cornea and extending a few mm on to the sclera. Left eye showed a single large dermoid covering temporal half of the cornea (almost 2/3rd of entire cornea) and extending a few mm onto the sclera.

Facial features

Hypoplasia of the right malar region, macrostomia, and he had multiple skin appendages over face on both sides, this was a unique feature which is not normally present in goldenhar syndrome.(Fig. 2,3)

Ears: The child had normal size but low set ears with bilateral pre-auricular tags and left sided pre-auricular pits.(Fig. 2,3)

His investigations revealed a normal haemogram, normal blood urea nitrogen and serum creatinine. Roentgenograms of the skull, spine, chest and abdomen were normal. Abdominal ultrasound and intravenous pyelogram revealed structurally and functionally normal kidneys.

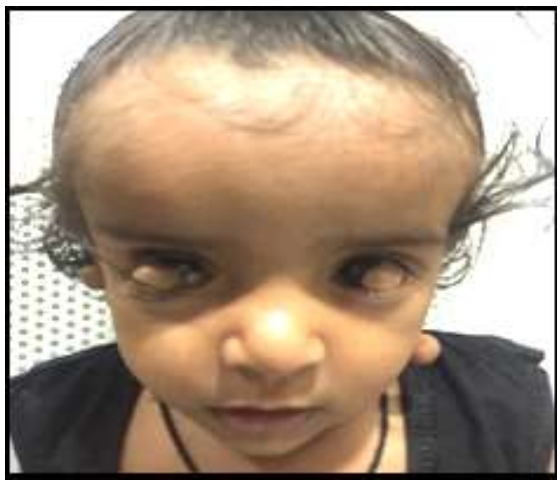

Fig. 1

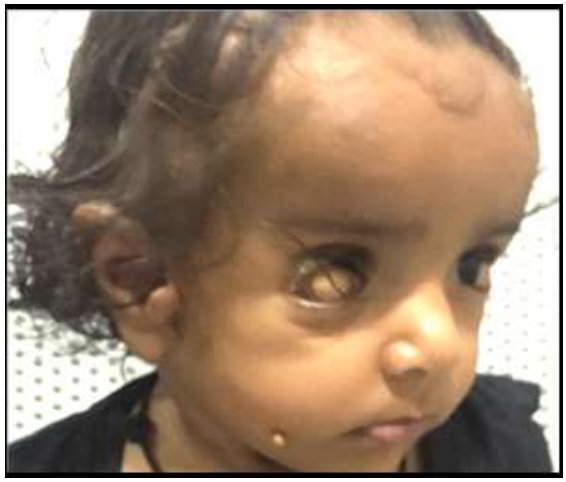

Fig. 2

Fig. 3

Fig. 4

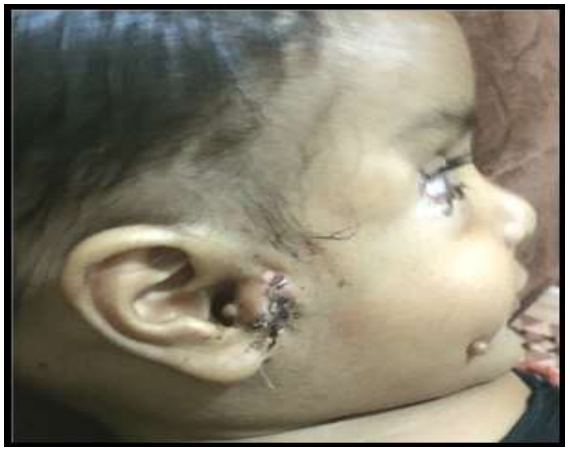

Fig. 5

Fig. 6

Management

In uncomplicated cases, the treatment of the syndrome varies with age and systemic associations and it is mainly cosmetic. The patient was taken for surgery under general anesthesia and both eyes were taken at the same time.

RE: both the epibulbar dermoids were removed by dissection from the cornea and sclera. The corneal involvement was upto 2/3rd of the cornea. The epibulbar dermoid was deeper than superficial layers of the cornea and sclera.(Fig. 4)

LE similar procedure was adapted in the left eye to remove the dermoid. (Fig. 4)

Simultaneously the facial and preauricular dermoids were excised. (Fig. 5,6)

Post-operative Management

The patient was kept on topical antibiotic eye drops, soft steroid drops and lubricant eye drops all three six times a day for two weeks. Since the post-operative corneal scar extended upto the pupil in the RE, the child was kept on 0.5% phenylephrine drops to keep the pupil dilated so as to prevent deprivation amblyopia orally the patient was on antibiotic syrup (Cefixime) BD and analgesic syrup BD dose empirically according to body weight for 1 week. The patient was followed daily for first week, then fortnightly for three months and then monthly. Post four months after surgery it was decided to do a an optical keratoplasty in the RE as the dermoid was deep and caused corneal thinning on excising it, also avoid amblyopia as the opacity was extending beyond the pupil as seen with the hand held slit lamp.

Discussion

Our case stands different that apart from the standard ocular and auricular pathologies multiple skin tags on face bilaterally has been rarely reported. The epibulbar dermoids also extended deep into the cornea, due to which Keratoplasty was our next step in managing the patient, to avoid amblyopia.

Further the patient's refraction shall be done and appropriate modality of refractive correction shall be prescribed, to prevent anisometropic amblyopia. Apart from treating the ocular epibulbar dermoids the preauricular and facial skin appendages were also surgically removed. The patient is still following up.

Differential Diagnosis

Other syndromes associated with multiple preauricular tragic include Treacher – Collins syndrome, Wolf – Hirsch horn syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome and Delleman syndrome. Teacher Collin syndrome is associated with maxillary and mandibular hypoplasia, but it is not associated with ocular and aural anomalies[3].

References

1. Riyaz A, Riyaz N. Goldenhar syndrome with unusual features. Indian J Dermatol Venereal Leprol. 1999;65:143–4.
2. Kulkarni V, Shah MD, Parikh A. Goldenhar syndrome: A case report. J Postgrad Med. 1985;31:177–9.
3. Reddy MV, Reddy PP, Usha Rani P, Hema Bindu L. Facio-auricular vertebral syndrome: A case report. Indian J Hum Genet. 2005;11:156–8.
4. Kapur R, Kapur R, Sheikh S, Jindal S, Kulkarni S. Hemi facial microsomal: A case report. J Indian Soc Pedod Prev Dent. 2008;26:34–40.
5. Miller TD, Metry D. Multiple accessory tragi as a clue to the diagnosis of the oculo-auriculo-vertebral (Goldenhar) syndrome. J Am Acad Dermatol.2004;50:11–3.