A case of Goldenhar-Gorlin syndrome with unusual association of hypoplastic thumb

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In Case 1, the hole was small initially and went unrecognized. The migration of a large bubble of PFCL into the vitreous cavity postoperatively on two occasions provided a clue to the entrapment of the fluid elsewhere. The choroidal hole became larger and prominent with increasing fibrosis. The fibrosis also prevented sclerochoroidal approximation in the vicinity of the hole, leading to persistent choroidal detachment in the area into which the silicone oil freely migrated.

In the second case, the hole was identified intraoperatively, only during the second surgery done six months after the injury. The hole presumably formed due to the traction by fibrosis that covered the bare sclera/RPE beyond the retinectomy edge, not an uncommon occurrence after large relaxing retinectomies. It is possible that the areas of choroidal atrophy and thinning can give way and progressively enlarge to form a choroidal hole in the presence of significant traction. Presence of silicone oil may not be a special concern but entrapment of PFCL could be problematic, since the surgeon is never sure as to the complete removal of the same at the conclusion of the surgery. Being heavier than water, PFCL gravitates down and could escape notice.

These two cases highlight an unreported occurrence of choroidal hole formation leading to suprachoroidal collection of tamponading agents such as PFCL and silicone oil. Both the cases also had hypotony caused possibly by severe fibrosis on the ciliary body. The combination of penetrating injury leading to choroidal damage and the significant proliferative vitreo-retinopathy that is associated with this situation can potentially lead to choroidal hole formation due to the severe traction. The presence of such a hole can complicate the surgical management of these eyes.

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Goldenhar-Gorlin syndrome manifests a number of craniofacial abnormalities that usually involve the face (hemifacial microsomia), eyes (epibulbar dermoid) and ears (microtia). It may also be associated with varying degrees of systemic and vertebral malformations. This syndrome presents at birth and the exact etiology is still unknown. The affections of the neural crest cells may have some role in the multiple malformations of the Goldenhar-Gorlin syndrome. The association of cardiac, pulmonary, central nervous system, renal and vertebral abnormalities have been described. Other skeletal malformations like extra ribs were reported. We report a rare case of Goldenhar-Gorlin syndrome with unique association of an isolated thumb hypoplasia.

Case Report

A seven-month-old male infant presented with fleshy masses in both eyes and facial deformity since birth. He was delivered normally at term by a 21-year-old mother. There was a history of taking multivitamin during her pregnancy but the exact nature and doses were not clear. His parents were non-consanguineous and belonged to the lower socioeconomic class. No other family members had such facial deformity or defect in the eyesight. On clinical examination the patient had right-sided hemifacial microsomia with malar hypoplasia, micrognathia and apparent macrostomia. The tongue was large and the palate was high arched. He had right lateral cleft lip with dental misalignment [Fig. 1]. Both ears were well formed with normal auditory functions but the right ear was slightly smaller (microtia) than the left one. Accessory preauricular appendages, two on each side were noticed in front of the auricle. A preauricular pit was found at a lower level near the angle of the mouth on both sides. Left-sided pit was surrounded by skin fold [Figs. 2, 3]. Both eyes had inferotemporal limbal dermoid which did not obstruct the pupil. Lipodermoids were present at the outer angle on each eye; the left one was larger and continuous with the corresponding dermoid [Figs. 2, 3]. Patient had right-sided hypoplastic thumb with flattened thenar muscles. It was shortened and dangled from the radial border of the hand resembling “floating” thumb [Fig. 4]. Both the forearms and arms were normal. Other systemic examinations revealed no abnormality.

Figure 1: The clinical photograph of the face showing left-sided hemifacial microsomia with right microtia and right lateral cleft lip with macrostomia

Figure 2: The clinical photograph of the left side of the face showing limbal dermoid, preauricular appendages and pit

Figure 3: The clinical photograph of the right side of the face showing limbal dermoid, preauricular appendages and pit

Figure 4: The clinical photograph of the hands showing right hypoplastic (floating) right thumb

Hemivertebra or butterfly vertebra defects were not found on complete spine films. X-ray of the right hand showed small first metacarpal (like phalanx) with deformed floating right thumb [Fig. 5]. Computed tomography (CT) scan of the brain, ECG and chest X-ray were within normal limits. The patient winced...
The syndrome presents at birth and its occurrence varies so much that actual modes of inheritance are only rarely documented. The present case was sporadic in nature. The sporadic nature can be better explained in the line of teratogenic effects of drugs on embryogenesis. In our case there was a history of taking multivitamins by the mother during pregnancy. Mounoud et al. reported a case of Goldenhar having a history of Vitamin A intoxication of the mother. Daily dose of 25000 IU of Vitamin A has teratogenic effects. That teratogen produces ill effects on neural crest cell formations which are essential for the formation of pharyngeal arches. Disruption of crest cells’ development results in variable craniofacial and ear malformation. Such crest cells appear to be vulnerable due to lack of superoxidase and catalase enzymes. Those enzymes are responsible for scavenging toxic free radicals.

Hyoplasia of the thumb is a type of distal form of ectromelia. Longitudinal or particle suppression of the limb buds results in ectromelia. The type and shape of the hand are regulated by HOX genes which are governed by the neural crest cells. So teratogenic effect of Vitamin A on neural crest cells’ formation may cause non-regulation of HOX genes that may produce characteristic features in this syndrome. The rare feature of this present case was the bilateral ocular involvement with epibulbar dermoid and extensive lipodermoid. After extensive MEDLINE search, we observed that the association of a thumb hypoplasia with the Goldenhar-Gorlin syndrome has not been reported so far.

Discussion

The Goldenhar syndrome was originally defined as a triad of congenital abnormalities consisting of epibulbar dermoid, preauricular appendages and pretagal fistulae. After Gorlin, the asymmetry of the face or hemifacial microsomia was also described as an usual association of this triad. Goldenhar-Gorlin syndrome is a congenital malformation of the structures derived from the first and second pharyngeal arches.

Hemifacial microsomia is a flattening of the face due to underdeveloped mandible (micognathia), maxilla and zygomatic bones with hypoplastic muscles for mastication and facial expression. The association of the lateral cleft lip with the hemifacial microsomia is not so common. The clefting of the lip usually occurs due to the failure of fusion between the maxillary and the medial nasal prominence.

The epibulbar limbal dermoid and lipodermoid are choristomas which are normal tissue in abnormal position. Bilateral limbal dermoids are rare in occurrence. Lipodermoids are fibrofatty tissue commonly located in the superotemporal epibulbar region and are generally bilateral. The larger one approaches the limbus and may blend with the adjoining dermoid.

Microtia involves defects of the auricle that range from absence of the ear (anotia) to small but well-formed ears. The external ear is developed from the hillocks on the first and second pharyngeal arches which are largely formed by neural crest cells. These cells play a role in most of the ear deformities. Appendages are abnormal developments of the accessory auricular hillocks whereas pith may indicate maldevelopment of the hillocks.

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Figure 5: X-ray of the right hand shows small first metacarpal (like phalanx) with floating right thumb