Hemifacial Microsomia and Accessory Auricles in an Adolescent Boy

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Sir,

Goldenhar syndrome is a rare disorder with developmental defects involving the first and second branchial arches being named in the Online Mendelian Inheritance of Man (OMIM) as “hemifacial microsomia” also known as “oculoauriculovertebral syndrome or spectrum.”

“Oculoauriculovertebral spectrum” was described by Goldenhar in 1952 with the triad of ear defects, epibulbar dermoids, and vertebral anomalies. [1]

Goldenhar-Gorlin syndrome is an expanded form of Goldenhar syndrome associated with facial clefting, mental retardation, vertebral abnormalities, cardiac or pulmonary anomalies, hemifacial microsomia involving hypoplasia of the maxilla, mandible, zygomatic bones,

A 17-year-old boy presented with multiple bilateral preauricular nodules since birth and swelling in the left eye which was progressively increasing in size, obscuring the vision. His hearing was normal. There was no maternal illness during pregnancy. On examination, he had epibulbar dermoid in the left eye [Figure 1a] and showed high-arched palate [Figure 1b] and multiple accessory auricles [Figure 1c and d]. Linear and rectangular atrophic macules were seen over cheeks [Figure 1c and d]. The atrophic macules appeared spontaneously without any history of inflammation or trauma. Flattening of the face was noted on the left side with micrognathia and macrostomia [Figure 2]. Audiogram was normal. X-ray and computed tomography scan of the cervical spine showed block vertebra between C5, C6 level [Figure 3a].

X-ray of the thoracolumbar spine showed hemivertebra at L5 level with left-sided lumbar scoliosis [Figure 3b]. There were no cardiac anomalies, and ultrasonogram of the abdomen was normal.
muscles of mastication, and facial expression.[2] Most cases of Goldenhar syndrome are sporadic.

The etiology is multifactorial with interaction between genetic and environmental factors such as exposure to chemicals or viruses during pregnancy and nutritional factors which may interfere with the development of vertebral and branchial systems.[3,4] Other manifestations of this syndrome include microtia, accessory auricles, preauricular pits or sinuses, lipodermoids, coloboma, microphthalmia, strabismus, failure of segmentation of vertebra, hemivertebra, cuneiform vertebra, and occipitalization of atlas.[5]

Other reported anomalies are club foot, congenital dislocation of the hip, Sprengel's deformity, radial limb defects, rib abnormalities, mandibular hypoplasia, cleft palate, occult spinal dysraphism, conductive or sensory neural hearing loss,[6] atrial or ventricular septal defects, coarctation of aorta, patent ductus arteriosus, and tetralogy of Fallot.[7]

The other syndromes resulting from defective development of the first and second branchial arches are Treacher Collins syndrome and Hallermann–Streiff syndrome.

The differential diagnoses for cystic eye lesions are Delleman syndrome and sebaceous nevus, central nervous system malformations, aplasia cutis congenita, limbal dermoid, and pigmented giant nevus with neurocutaneous melanosis syndrome.

Accessory auricles and preauricular sinuses occur with Treacher Collins, Goldenhar, Nager's acrofacial dysostosis, 4p-, oculoauriculovertebral, Kabuki’s, Townes-Brocks, Haberland, and branchio-oto-renal syndromes.

Our patient presented with classical features of Goldenhar syndrome, for whom multidisciplinary surgical corrections and regular follow-up are being planned. There are about 3% chances of passing it on to the offspring. Goldenhar syndrome support system can help patients and parents. Atrophic macules in Goldenhar syndrome have not been reported in the literature so far.

Whether the atrophic macules over the face were due to atrophy maculosa varioliformis cutis or an association with Goldenhar syndrome is not known.

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.

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