Case Report

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Goldenhar Syndrome with Radial Club Hand: An Unusual Presentation

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Abstract

Goldenhar syndrome also known as Facioauriculovertebral sequence is a rare congenital condition presenting with the asymmetry of the face, under developed ear, cleft lip/palate and abnormalities of the spine. In addition, Goldenhar can affect the heart, kidney, lungs and limb abnormalities. We would like to share one such case presented to us with a constellation of features of Goldenhar syndrome along with a contralateral radial club hand.

Keywords: Goldenhar syndrome; Hemi facial microsomia; Radial Club hand; Radial deficiency.

Introduction

Goldenhar syndrome also known as Facioauriculovertebral sequence, oculo-auriculo-vertebral (OAV) syndrome was first described by Dr. Maurice Goldenhar in 1952 as a triad of epibulbardermoids, preauricular appendages and pre tragal fistulae.[1] Since Goldenhar's original description, other manifestations have been added, including vertebral abnormalities, upper eyelid colobomas, subconjunctival lipomas, ear anomalies, hearing loss, unilateral Facial hypoplasia, micrognathia, cleft or high-arched palates, and congenital cardiac anomalies.[2]

The incidence has been reported to be 1:3500-1:5600 with a male to female ratio of 3:2.3.

The condition is sporadic in 90% of cases 4 and positive family histories have been reported (10%) suggesting autosomal dominant or recessive inheritance.

The underlying cause of Goldenhar disease is poorly understood, the pathophysiology is believed to be occlusion of the stapedial artery during organogenesis affecting structures that originate from the first and second branchial arch.[5]

Radial aplasia or hypoplasia is another rare congenital abnormality with an estimated live birth incidence of 1:5000. In addition to an abnormal radius and/or ulna, there may also be anomalies of the humerus, scaphoid, trapezium, metacarpals, and thumbs as part of a wide spectrum called 'radial ray malformations.[6] From the late 1800s, the varied etiology of congenital malformations has been observed, with bilateral forearm abnormalities being more often associated with an underlying genetic etiology than unilateral lesions.[7]

Case Report

A 6-year-old boy reported with the complaint of deformity of the left wrist. The patient was born to second-degree consanguineous parents and his younger sibling is normal. History revealed pregnancy and birth were uneventful. Speech milestones were delayed. The performance in school was satisfactory. On examination, the patient had facial asymmetry, hypoplasia of the mandible,

and retrusion of the midface with loss of malar prominence on the right side. The. The angle of the mouth was deviated to left. Low facial nerve palsy. (Fig. 1) The base of the nose was broad, low set underdeveloped right ear with atresia of external auditory meatus with preauricular tags. (Fig. 2)

Intraoral examination revealed a scarred soft palate (operated for cleft of the soft palate at the age of 10months) and Angle's class III malocclusion with crowding of both maxillary and mandibular anterior teeth and anterior open bite. The child had chronic generalized gingivitis associated with poor oral hygiene.

The patient had type 4 left radial club hand with a hypo plastic thumb with contracted first web space and limited movements of fingers. (Fig. 3) The patient was also noted to have hypo plastic ribs on the left side with poorly developed pectoralis muscles. (Fig. 4)

The patient had thoracolumbar scoliosis and webbing of the neck (Fig. 5) Cardiac evaluation, eye examination was normal.

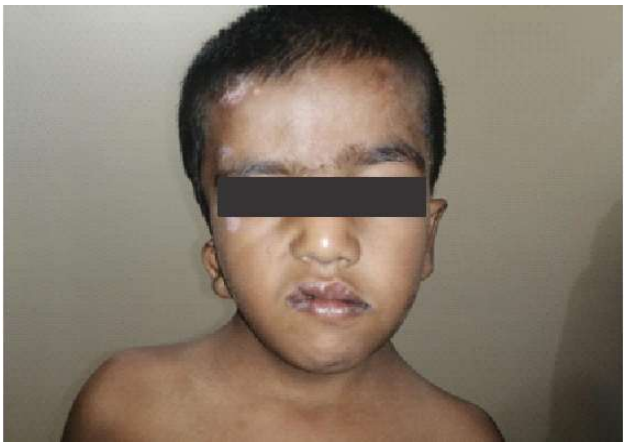

Fig. 1: Facial asymmetry with facial nerve involvement

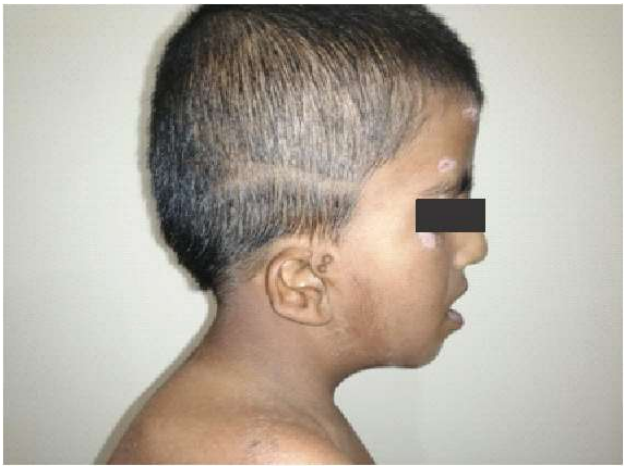

Fig. 2: Low set ear with preauricular tag

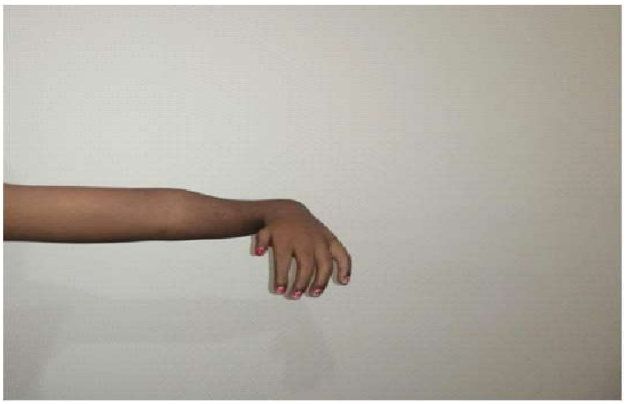

Fig. 3: Type 4 Radial Club hand

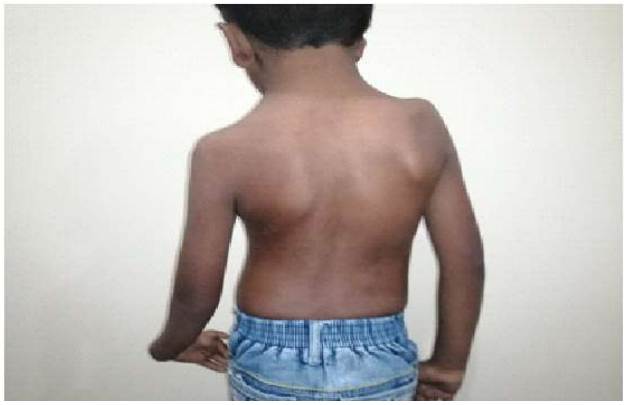

Fig. 4: Hypoplastic left hemithorax

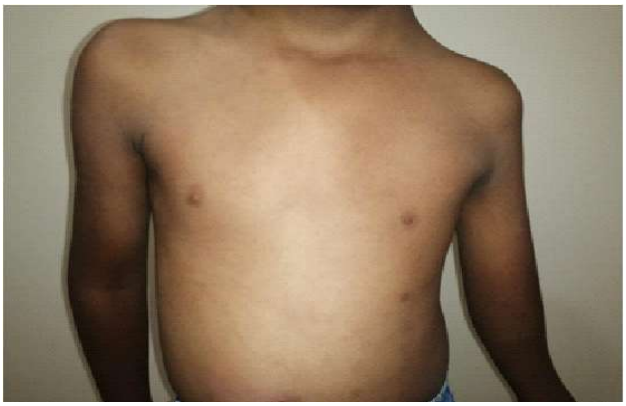

Fig. 5: Scoliosis with webbing of the neck

Discussion

The criteria for the diagnosis of Goldenhar's syndrome consisted of an eye abnormality (lipoma, lipodermoid, epibulbar dermoid, or upper eyelid coloboma) associated with the ear, mandibular, or vertebral anomalies (two of the three).

It has been estimated that epibulbardermoids occur unilaterally in one-third of patients with Goldenhar' ssyndrome,. review of the literature suggests, 23% were bilateral, 53% were unilateral,

and 24% had no epibulbar dermoid.[8]

Goldenhar's syndrome representsthe spectrum of Hemifacial Microsomia (HM) which is predominantly unilateral malformation of craniofacial structures that developed from the first and second branchial arches. However, both HM and the Goldenhar syndrome may represent gradations in the severity of a similar error in morphogenesis.[7]

Various other anomalies associated are.[1] Cardiovascular system: Tetralogy of Fallot. Dextrocardia, transposition of great vessels, right bundle branch block, pulmonary stenosis, atrial septal defect.[2] Central nervous system anomalies hydrocephalus, meningoencephalocele, mental retardation.[3] Others include renal agenesis, inguinal hernia, hemangiomas, rectovaginal fistula, and club feet.[3]

Radial club hand is a rare association with Goldenhar's syndrome. The most common anomalies observed were hypoplasia of thumb and hypoplasia/ agenesis of the radius[9] significantly associated with short stature.[10]

Radial ray deficiency with goldenhar syndrome is a rare association, our case in addition to the existing number.

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