Case Report

Atypical Presentation of Goldenhar Syndrome

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Shubhrakanti Sen,[1] Debmalya Maity,[2] Arnab Koley[2]

ABSTRACT

Introduction

In 1952 Goldenhar described a case with triad of pre auricular tags, mandibular hypoplasia and ocular (epibulbar) dermoid and described the case as Goldenhar Syndrome.

Case Report

A case of Goldenhar Syndrome without ocular involvement is presented.

Discussion

Goldenhar syndrome is also known as oculoauriculovertebral dysplasia due to presence of additional vertebral anomalies. Exact etiology of this disease is not known. Most of the cases are sporadic, though autosomal recessive/dominant and multifactorial inheritance has also been suggested. Chromosomal analysis shows no abnormalities.

Keywords

Goldenhar Syndrome

Goldenhar syndrome is a rare disorder with an incidence of 1 in 5800 live births with male: female ratio 3:2. It is presumed to be an inherited condition causing morphological abnormalities of the parts developed from the first and second branchial arch during blastogenesis.[1] Goldenhar first described the case in 1952 as a disease that presents as a combination of several anomalies such as epibulbar dermoids, preauriclar appendices and malformation of ears. It is also referred as oculo-auriculo-vertebral (OAV) dysplasia and hemi facial microsomia. Most of the cases are sporadic, though autosomal recessive/dominant and multifactorial inheritance has also been suggested. Chromosomal analysis shows no abnormalities.[2]

1 - Department of ENT, WBMES NRS Medical College, Kolkata

2 - Department of Gyneacology and Obstetrics, Nil Ratan Sirkar Medical College & Hospital, Kolkata

Corresponding author:

Dr Shubhrakanti Sen

email: sen.shubhrakanti@gmail.com

Some associations recorded in the literature are microstomia, micrognathia, high arched palate, cleft palate, bifid tongue, malocclusion and other dental anomalies.[3] Some authors also pointed out facial muscle hypoplasia, vertebral anomalies, eye anomalies,[1] and disorders of central nervous system, visceral anomalies,[4] cardiovascular[5] and genitourinary abnormalities.[6] The presence of anomalies of the ears and limbs are necessary for the diagnosis of this syndrome.

In this article we are presenting an interesting case of neonate diagnosed as Goldenhar syndrome without ocular involvement.

Case Report

A 2.5 Kg, male preterm baby (gestational age 36 weeks) delivered by elective caesarean section (indication: fetal distress), cried immediately after birth & transferred to NICU (neonatal intensive care unit) with multiple congenital anomalies.

Antenatal history: A 26 years old, primigravida with a history of intake of oral abortificiant (Levonorgestrel - 1.5

Fig. 1. Showing Right ear-grade 2 microtia, preauricular tag, sinus and pit

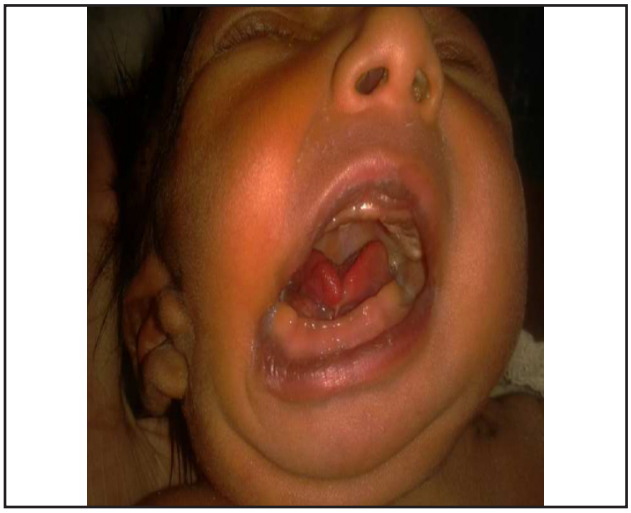

Fig. 2. Macrostomia, tongue tie, bifid tongue, right sided deviation of the angle of mouth.

mg) during her first trimester as per advised by medical practitioner. But as the abortion did not occur, mother continued the pregnancy. She had irregular antenatal checkups. First antenatal ultrasonography done on 23rd week of gestation, revealed major congenital anomalies. Mother was non smoker and non alcoholic without any significant illness.

Neonatal period: Baby had feeding difficulty without any jaundice or convulsion.

Clinical examination: Baby had dimorphic facies, vitals - stable, cardinal clinical signs – normal.

Head to foot examination:

Head Circumference- 33cm, anterior fontanel –normal, Chest circumference- 30cm, Length-48cm

Ears

Right ear-grade 2 microtia, preauricular tag, sinus and pit. (Fig. 1) Left ear-normal

Nose: normal

Oral Cavity: macrostomia, tongue tie, bifid tongue, right sided deviation of angle of mouth with bifurcation of lower lip at right angle of mouth.

Face: Micrognathia.

Neck: Short neck.

Eye: No obvious anomaly seen.

Upper Limb: Right upper limb deformed with short forearm, hypoplastic right thumb & radial deviation of hand, fixed flexion deformity of elbow joint. (Fig. 2)

Chest, Abdomen and Genitalia: Normal.

Lower Limb: No obvious deformity

Investigations:

Complete Blood Count: Normal.

Echocardiography: Small PDA with left to right shunt, osteum secondum type ASD.

Ultrasonography: Hypoplasia of right parotid gland & irregularity in right half of mandible seen. No abnormality of abdominal organ seen.

USG brain: Normal.

Skiagram: Hypoplastic right half of mandible, absent radius in right forearm. (Fig.3) Lower cervical vertebral body (C6, C7) was fused and bifid left 3rd & 4th rib were seen. (Fig.4)

Karyotyping: Normal.

Discussion

The physical findings of the baby had some similarities with Treacher Collins Syndrome which is characterized by bilateral affection without any aural or ocular abnormality.[7]

But the above finding of the baby clinched us to

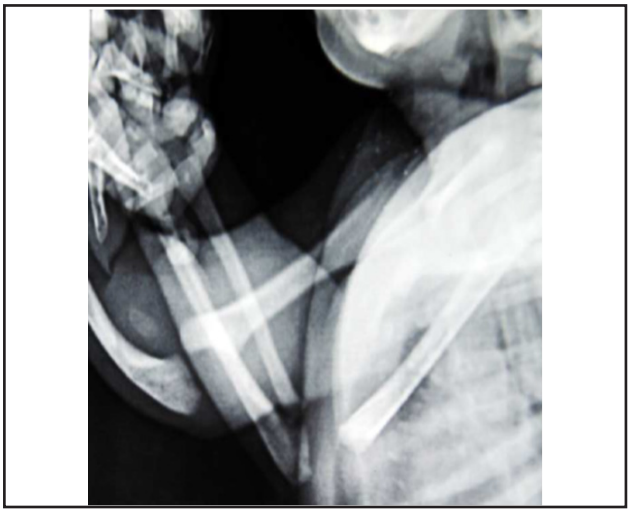

Fig. 3. Skiagram showing absent radius in right forearm

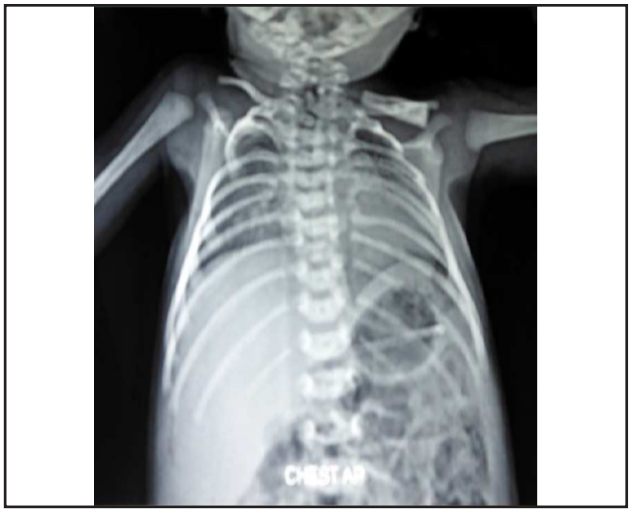

Fig. 4. Skiagram showing lower cervical vertebral body (C6, C7) was fused and bifid left 3rd and 4th rib

the diagnosis of Goldenhar syndrome. Hemifacial microsomia or Goldenhar syndrome manifests itself in degrees ranging from nearly unnoticeable to extremely severe. The classic symptoms of Hemifacial microsomia include: underdevelopment of the jaw on one side (micrognathia), underdeveloped cheek bone on the affected side, underdeveloped or deformed an outer ear (microtia) and a missing or undersized ear canal (congenital aural atresia).

Additional symptoms of Goldenhar syndrome include: anomalies of the spine (most typically cervical vertebrae deformities), narrowing of one eye, a soft white or yellow nodule located in the eye (epibulbar dermoids) and notched eyelids.

Most individuals with Goldenhar syndrome or Hemifacial microsomia have a malformed outer ear, a condition called microtia. In approximately one third of cases of Goldenhar syndrome, the microtia is bilateral. Microtia is rated on a four-point scale. In Grade 1 microtia, the ears look almost normal but are smaller than average. Grade 2 microtia is characterized by having a curved mass of tissue rather than a formed outer ear. Grade 3, which is the most common form of microtia, consists of having only small bumps of skin. Grade 4, sometimes referred to as "anotia", consists of having no external ear at all.[8] In this child it was grade 2 microtia.

The abnormalities are found to be unilateral in 85% of cases and bilateral in about 10-33% cases.[9] In asymmetric involvement right side is more affected than left side.*10* Our reported case had similar involvements. The reported frequency of cardiovascular abnormality ranging from 5-58%. This baby also had cardiac lesion in the form of ASD & PDA. Some association was found between maternal intake of some drugs like retinoic acid, thalidomide etc with development of Goldenhar syndrome.[11] Our case had a history of abortificiant drug (Levonorgestrel - 1.5 mg) intake in first trimester whose association with this syndrome was not reported earlier.

In Goldenhar's syndrome ocular anomalies especially bilateral dermoid present in 60% of cases and vertebral and ear abnormalities are present in 40% cases.[12] The characteristic vertebral, ear, cardiac, facial, rib, Mandibular anomalies all were present in our case. But surprisingly we didn't find any ocular abnormalities, which were reported in maximum cases in literature. So the salient features of the patient presented here are:

I. May present without ocular involvement.

II. Association with intake of abortificiant.

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