Goldenhar Syndrome - A Case Report

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INTRODUCTION:

Goldenhar syndrome also known as oculo-auriculo-vertebral syndrome (OAVS)[1], hemifacial microsomia, facioauriculovertebral syndrome, Goldenhar-Gorlin syndrome, first & second branchial arch syndrome, & oculo-auriculo-vertebral dysplasia is characterised by triad of ocular, auricular & vertebral anomalies. It was first recorded by German physician, Carl Ferdinand Von Arlt, in 1845. In 1952, Maurice Goldenhar described a patient with a triad of accessory tragi, mandibular hypoplasia & ocular (epibulbar) dermoids & called this constellation of features as Goldenhar Syndrome[6]. In 1963, Gorlin and Pindborg named this syndrome as oculoauriculovertebral dysplasia due to presence of additional vertebral anomalies. In early 1990's this condition was better understood & it was agreed that, besides the picture described by Goldenhar (1952) & Gorlin (1963), this syndrome may present heart diseases & hypoplasia of the zygomatic, mandibular & maxillary bones. Due to craniofacial malformations, an affected individual's face may appear smaller on one side than the other (hemifacial microsomia). We have a rare case of Goldenhar syndrome with a unique swelling in the right molar region.

CASE REPORT:

An 18 month old female child, born to non-consanguineous marriage, presented with complaints of asymptomatic swelling on the right side of the cheek since birth and intraoral swelling in upper right back tooth region with tooth erupting from the sweiling since 1 month. There was no increase in the size of the lesions. The child was born of a full-term, normal delivery and there was no history of any maternal illness during the pregnancy. The family history was non-contributory.

On extraoral examination, asymmetry of the face was noted with the swelling on right side of the face, 3cm in diameter, extending vertically from lateral canthus of the eye to the angle of the mouth. Corner of mouth was extended on the right side (macrostomia).

Scar mark was present along the line joining the tragus and the angle of the mouth with the history of surgical excision of the tissue tag for the esthetic reason. Examination of the oral cavity revealed a swelling in upper right back molar region and a tooth was erupting through that swelling. Swelling was approximately 1.5 cm in diameter and bony hard in consistency. The swelling along with the tooth was displaced distally and buccaly.

Ocular examination showed decreased width of palpable fissure of the right eye (micropthalmia). Skeletal examination was clinically normal. Examination of the cardiovascular, respiratory, gastrointestinal and genitourinary systems revealed no abnormality.

The radiographic examination showed an accessory alveolus along with an erupting tooth. The tooth was abnormal in shape.

The condition was diagnosed as a case of Goldenhar syndrome based on accessory tragi, macrostomia, microotia. She was counseled regarding the condition.

DISCUSSION :

Goldenhar syndrome is a rare developmental anomaly featuring the triad of ocular, auricular & vertebral anomalies. This syndrome involves structures arising from the first & the second branchial arches. The incidence of Goldenhar syndrome has been reported to be between 1:3000 to 1:5000 with a male to female ratio of 3:21. Generally, right side of the face, body or both is affected more commonly and severely than the left side.

The exact aetiology is not known. However, it is possible that abnormal embryonic vascular supply, disrupted mesodermal migration or some other factors lead to defective formation of the branchial and vertebral system. It may represent a defect in blastogenesis that could be attributed to interference in cephalic neural crest cell migration. A new theory put forward for this developmental anomaly states that OAVS occurs because of ectodermal nondisjunction early in embryonic development during 4th to 8th weeks of intrauterine life with subsequent mesodermal tethering[2]. Most cases have been sporadic. Autosomal dominant, autosomal recessive & multifactorial modes of inheritance have also been suggested. There is no confirmed genetic component despite of reported cases of autosomal dominant pattern & trisomy 8, 9 & 22 occurrences.

The ingestion of some drugs such as cocaine, thalidomide, retinoic acid & tamoxifen by mother was also related with development of the disease[1]. The maternal diabetes also was suggested as etiologic factor. In our case, there was no history of maternal drug intake, any febrile illness or maternal diabetes during pregnancy.

The diagnosis of Goldenhar syndrome mainly is based on the clinical aspect & is associated with both systemic condition & radiologic findings. The cardinal feature of Goldenhar syndrome includes ocular, auricular vertebral & craniofacial anomalies.

Ocular changes include microphthalmia (abnormal smallness of eyes), narrowing of the eyefolds (palpebral fissures) between upper & lower eyelids (blephrophimosis), epibulbar dermoids, lipodermoids and coloboma. Aural features include preauricular tragi, hearing loss and microtia; and vertebral anomalies such as spins bifida occulta, scoliosis, hemivertebrae and cervical fusion. The abnormalities are found to be unilateral in 85% of cases and bilateral in 10-33% cases.

Intraoral structures are also affected in this condition. The findings that can be observed are malocclusion[4], tooth discrepancies, macrostomy caused by underdeveloped lower jaw, agenesis of third molars & second premolars on the affected sides, enamel & dentin malformations & delay in tooth development. Moreover, patients with hemifacial microsomia are often present with asymmetric development of masticatory muscles as well as agenesis of salivary glands & rarely palatal clefts[4].

Additional malformations seen with this syndrome are, cardiac anomalies (5-58%)[5] such as Tetralogy of Fallot and ventricular septal defects, arterial septal defects. Urologic anomalies, which include ectopic kidneys, renal agenesis, urethral duplication, multicystic dysplastic kidneys. Genital anomalies like hypoplastic vagina, hypospadias & scrotal anomalies. Central system anomalies like encephalocele, hydrocephaly, agenesis of corpus callosum, calcification of falx cerebri, hypoplasia of septum pallicidum & memingocele. Vascular system anomalies, gastroinstinal anomalies & pulmonary anomalies may also occur.

Other syndromes associated with multiple preauricular tragi include Treacher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager's acrofacial dysostosis, Wildervanck syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome and Delleman syndrome[3]. Treacher Collins syndrome is associated with maxillary and mandibular hypoplasia but is not associated with ocular and aural anomalies.

The treatment of the disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases. In patients with mandibular hypoplasia, reconstruction can be done with rib grafts and a bone distraction device can lengthen an underdeveloped maxilla. External ear reconstruction surgeries can be performed at the age of 6 to 8 years. In patients with milder involvement, jaw reconstruction surgeries can be done in the early teens; epibulbar dermoids should be surgically excised. Structural anomalies of the eyes and ears can be corrected by plastic surgery. Prognosis of the disease is good in otherwise uncomplicated cases without any systemic associations.

Fig 1- Frontal view of the face with asymmetry on right side of the face

Fig 2- Scar marks of the excised tissue tags on right side of the cheek

Fig 3- Intraoral view of the accessory alveolus along with erupting tooth

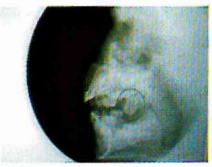

Fig 4- Radiographic lateral view

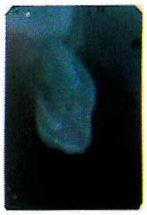

Fig 5- Intraoral periapical radiograph of the involved tooth

REFERANCES

1. Amitava D, Biswarup R, Debabrata D, Somnath D. A case of Goldenhar-Gorlin syndrome with unusual association of hypoplastic thumb. Indian J Ophthalmol 2008;56:150-
2. Bijal M., Chitra N., Shankar S., Sangeeta A. Goldenhar syndrome with unusual features. Indian J Dermatol Venereol Leprol 2008;74(3):254-256.
3. Mahato SK, Grover SB. Oculo-auriculo-vertebral dysplasia (OAVD): A case report. Indian J Radiol Imaging 2003;13:456-457
4. Ahmet G, Yasemin K, Osman T, Hakan G, Perçem B, Serap k. Goldenhar's syndrome with lens subluxation; a case Report. Case Rep Clin Pract Rev 2007; 8: 69-72
5. David Healey, Merv Letts, James G. Jarvis. Cervical spine instability in children with Goldenhar's syndrome. Can J Surg 2002;45(5):341-344.
6. Jaison SG, Batra N. Goldenhar syndrome with multiple additional anomalies. Indian J Ophthalmol 1996;44:42-4