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Goldenhar Syndrome - A Rare Entity

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Abstract: ***Goldenhar syndrome is a rare congenital anomaly which was first described by Goldenhar in 1952. Goldenhar syndrome is also known as oculo-auriculo-vertebral (OAV) dysplasia. This term oculo-auriculo-vertebral dysplasia was described by Gorlin, Cohen and Levine consisting of spectrum of anomalies ranging from hemifacial microsomia (HFM) to Goldenhar syndrome which also includes epibulbar dermoids and vertebral anomalies. The exact cause of Goldenhar syndrome is unknown however etiology behind this syndrome is considered to be multi-factorial pattern, i. e. combination of gene interactions and environmental factors that affects the development of the first and second pharyngeal arches during the first trimester of pregnancy. We reported a patient with this complex, rare congenital anomaly in our radiology department in which all classical findings of goldenhar syndrome were present. Findings of this syndrome are presented here.***

Keywords: Oculo-Auriculo-Vertebral, OAV, syndrome, Goldenhar-Gorlin syndrome

1. Case Report

A twenty two years old male patient came to our department of radiology with clinical history of deviation of mouth towards right side. Patient was not suffering from any systemic disease and his intelligence quotient was normal. He was advised CT scan of face and temporal bone by clinician. CT scan of patient revealed facial asymmetry with hypoplastic right side of face, elongation of right angle of mouth and deviation of chin towards right side. Hypoplasia of right zygoma, maxilla and right hemi-mandible was noted more so involving right mandibular ramus and mandibular condyle. In addition, deformities were noted in right ear in form of right microtia with small deformed right pinna. Right external auditory canal and middle air cavity were not formed, instead these cavities were replaced by thick bony tissue. Right middle ear ossicleswere completely absent, however right inner ear and internal auditory canal structures were normal. Left ear was also normal.

Deformities were also noted in cervical spine in form of hypoplastic C1 vertebrae, rotational deformity at C1-C2 level with rotation of C2 right anteriorly in relation to C1 and fused lateral masses of C1 and C2 on left side. Also there was fusion of body and posterior element of C2-C3 and C6-C7 vertebrae s/o fused vertebrae.

Basilar invagination was noted in form of odontoid process tip lying 6.5 mm above Chamberlain line (horizontal line drawn from posterior aspect of hard palate to opisthion).

Based on radiological findings discussed above, we made a diagnosis of Goldenhar Syndrome.

2. Discussion

Facio-auriculo-vertebral syndrome was first described by Canton in 1861 and by German physician Von Arlt in 1881 [1]. Later on triad of epibulbardermoids, auricular appendages and mandibular hypoplasia was described by Goldenhar and named it as Goldenhar syndrome [2]. Gorlin, Cohen and Levine described it as oculo-auriculo-vertebral (OAV) dysplasia due to presence of additional vertebral anomalies [3]. Goldenhar syndrome is a rare complex congenital anomaly and the exact cause of Goldenhar syndrome is unknown but considered to be multifactorial, i.e. a combination of gene interactions and environmental factors that affects the development of the first and second branchial arches during the first trimester of pregnancy [4]. Most of cases reported are sporadic, however autosomal dominant, autosomal recessive and multifactorial modes of inheritance have also been suggested. In 50% of patients with Goldenhar syndrome systemic features can be found. Cardiovascular anomalies predominantly ventricular septal defects and tetralogy of fallot are associated with this syndrome [5]. Apart from these cleft lip and palate, webbed and short neck, inguinal and umbilical hernia may be associated.

Etiology

Multifactorial pattern of inheritance i. e. combination of genetic and environmental factors is linked to development of Goldenhar syndrome. In environmental factors intake of teratogenic drugs such as cocaine, thalidomide, retinoic acid, tamoxifen especially in first trimester of pregnancy leads to the development of this complex anomaly [6, 7]. Maternal diabetes is also considered as one of important risk factor for development of this anomaly [8]. Vitamin A intoxication produces teratogenic effect in form of disturbance in formation of pharyngeal arches by attacking neural crest cells [9]. Heavy alcohol consumption during pregnancy is also linked to this syndrome [10].

Differential diagnosis

One of major differential diagnosis of Goldenhar syndrome is Treacher Collins Syndrome consists of mandibular and maxillary hypoplasia, but ocular and aural anomalies are not present in this syndrome [11]. Other differential include Wildervanck syndrome (cervico-ocular-acoustic dysplasia).

Management

As patients of Goldenhar syndrome suffer from facial asymmetry and other ocular & aural anomalies, therefore cosmetic surgery plays very crucial role in management of these patients. Mandibular hypoplasia is treated by reconstruction of mandible with rib bone grafts and maxillary hypoplasia is treated by using distraction techniques. Reconstruction of external ear can be done particularly at age of 7-8 years. Epibulbardermoids should be excised. Apart from these surgical treatments discussed, proper counseling of parents about this syndrome plays very

important role. Prognosis is generally good if no systemic disease is associated [11].

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