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Case report

GOLDENHAR SYNDROME: A CASE REPORT WITH FACIAL OCULAR AURICULAR VERTEBRAL MANIFESTATIONS

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ABSTRACT

Goldenhar's Syndrome (GS) is a rare condition described initially by Von Arlt. It is characterized by a combination of anomalies: epibulbar tumors, preauricular tags and malformation of the ears. In 1963, Gorlin suggested the name oculo-auriculo-vertebral (OAV) dysplasia for this condition and also included vertebral anomalies as signs of the syndrome. The malformations are said to arise from the first and second branchial arches. This work reports a case of GS in 11-years-old boy with the clinical signs of pseudo macrostomia, preauricular tags and epibulbar tumours.

Keywords: Epibulbar Tumours, Goldenhar Syndrome, Preauricular Tags, Macrostomia.

INTRODUCTION

There are several synonyms of Goldenhar Syndrome, which are Facio-Auriculo-Vertebral Sequence (FAVS), Oculo-Auricle-Vertebral spectrum (OAVS) and First and Second Branchial Arch Syndrome. GS is caused by both genetic and environmental factors. It is often referred to as "Hemifacial Microsomia" when it primarily involves the jaw, mouth and ear on one side of the body.[1] The characteristic asymmetrical malformations of the face, eye and ear were first recorded by the German physician Von Arlt in the 19th century but in 1952 a French Ophthalmologist, Goldehnar Maurice reviewed the subjects and described the condition as a combination of several anomalies such as Dermal / Epibulbar tumors, Preauricular Appendages and Malformed Ears.[2] The malformations of this syndrome arise from defects in the 1st and 2nd branchial arches.[3] Currently no genetic/DNA test is available for GS; hence prenatal diagnosis and treatment is not possible.[4] The prevalence of this condition ranges from 1:3500 to 1:5600 live births with a male to female ratio of 3:2.[5]

CASE REPORT

An 11-years-old boy presented with multiple decayed teeth to SDM College of Dental Sciences and Hospital, Dharwad. Extra oral examination showed a crease over the left lateral commissure of the mouth, which was surgically treated and epibulbar tumors of both the eyes for which he underwent two surgeries till date (Fig 1a and 1b).

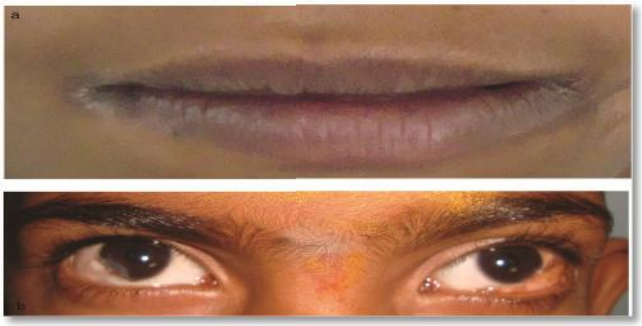

Fig 1: (a) lateral crease on the left side of the mouth (b) epibulbar tumors in both the eyes.

He had a convex profile. The left external helix was anteriorly rotated compared to right ear with a surgical scar in the preauricular region indicative of auricular tag removal. A pit was present in front of right tragus (Fig 2a, 2b and 2c). On examination showed macrostomia approximately 45mm with a mild deviation of the mandible to the left side and had mixed dentition with anterior open bite. Patient also had multiple carious teeth (Fig 3).

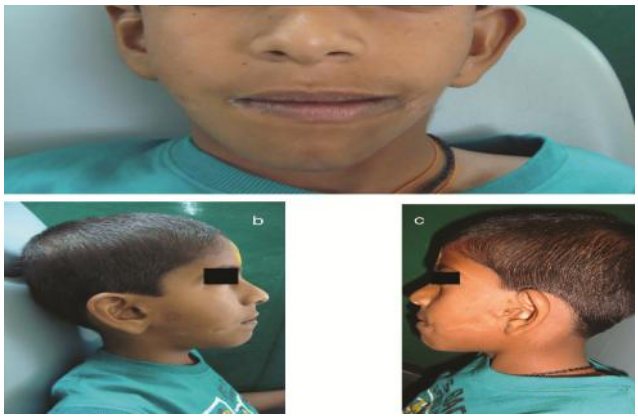

Fig 2: a) anterior rotation of the left external helix (b) preauricular pit and surgical scars (c) accessory tragi and surgical scar of left lateral facial cleft.

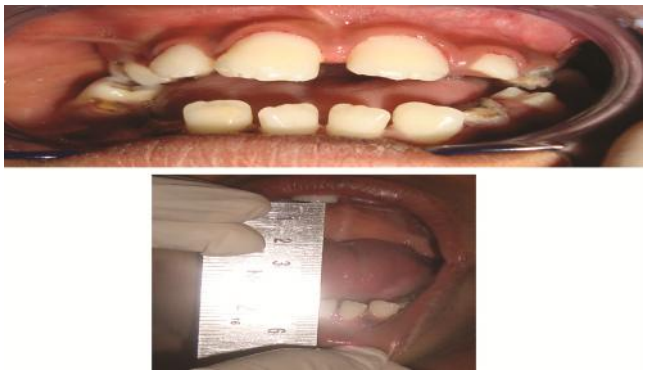

Fig 3: Anterior open bite and macrostomia

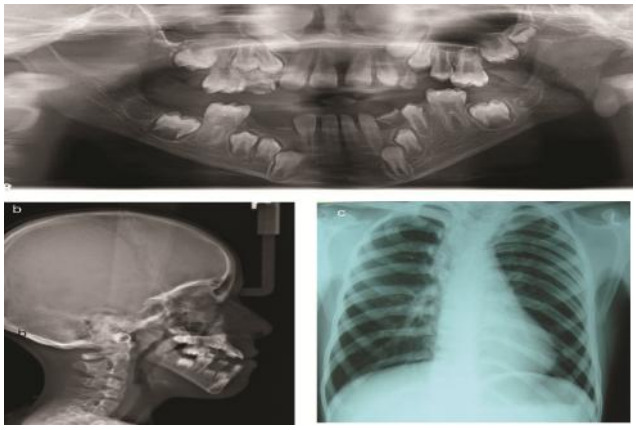

Fig 4: (a) Digital OPG showing hypoplastic condyle and coronoid processes and flattening of glenoid fossa and articular eminence (b) lateral cephalograph shows block vertebrae with C2 and C3 (c) Chest radiograph reveals crowding of ribs on the left side and scoliosis of vertebral column

His parents were non-consanguineous and there was no family history of similar defects or any maternal illness/medication during pregnancy. There was no evidence of any psychological disability.

Radiological examination: Digital Orthopantamo graph (OPG) revealed hypoplastic condyle and coronoid processes with flat glenoid fossa and articular eminence. Lateral Cephalograph revealed block vertebrae with C2 and C3. Chest radiograph showed crowds of ribs on the left side and scoliosis of vertebral column (Fig 4a, 4b and 4c).

DISCUSSION

Majority of this syndrome occur sporadically and there is a very small chance of familial occurrence. Although exact etiology of Goldenhar syndrome is unknown, autosomal recessive or dominant inheritance has been suggested.[6] Multifactorial inheritance due to interaction of many genes, possibly in combination with environmental factors such as chemical exposure can be the etiology. The diagnosis of Goldenhar syndrome is mainly based on clinical aspect and associated radiologic findings and systemic conditions.

Varied clinical features of syndrome

1. Ear: Preauricular skin tags, dysmorphic ear, conductive hearing loss.
2. Neck: Branchial fistula, webbing, abnormalities of sternocleidomastoid muscle.
3. Abdominal wall: Divarication of recti, umbilical hernia, inguinal hernia
4. Eye: Epicanthal folds, upper / lower lid coloboma, epibulbar dermoids, epiphora, microopthalmia
5. Face: Hypoplasia of maxillary, zygomatic, coronoid and condylar process, macrostomia, mandibular ramus asymmetry.
6. Back: Pilonidal dimple, kyphoscoliosis.
7. Hands / Fingers: Polydactyly, clinodactyly, single palmar crease.
8. Skeletal: Cervical fusion, hemivertebrae, and scoliosis.
9. Cardiovascular: Cardiomegaly, ASD, VSD, TOF.[7-10]

Our patient had macrostomia (surgically treated), an accessory tragi on the left side, preauricular pit on the right side, preauricular skin tags and epibulbar tumors on both the sides (surgically treated), hypoplasia of the condyle and coronoid processes with flat glenoid fossa and articular eminence on the left side, block vertebrae (C2 and C3), crowding of ribs on the left side and scoliosis of the vertebral column.

CNS: The frequency of intellectual retardation varies from 10-25% from a practical standpoint; those children with encephalocele, severe microphthalmia or with malformations not traditionally seen in Goldenhar syndrome have a severe risk of mental retardation

Prognosis: Prognosis is variable and depends on the presence and severity of associated cardiovascular, neurological and other complications. The life span of children is normal.

In our patient, the prognosis would have been better.

Our patient had undergone plastic surgery for aesthetic correction. Specialized dental care was advised for carious teeth and malocclusion. Other treatment options in GS include grafting to correct the cheekbones, jaw surgery, eye surgery, ear reconstruction, staged orthodontics, palatal closure, speech therapy (cleft palate cases) and hearing aids. The differential diagnosis includes Treacher Collins syndrome (TCS) that affects both sides of face symmetrically and lacks ocular and vertebral malformations of GS. Pierre Robin syndrome always consists of cleft palate, micrognathia and glossoptosis. Moebius syndrome is a nonfamilial deficient development of cranial muscles consisting of facial diplegia with bilateral paralysis of the ocular muscles, particularly those supplied by abducens. Other birth defects similar to GS are Nager Acrofacial Dysostosis, Maxillofacial dysostosis, Townes-Brocks Syndrome and Brachio-oto-renal syndrome. There is no vaccine available to prevent GS in the present date.

CONCLUSION

The effect of Goldenhar syndrome is more evident as the child grows, because of delay in the growth and development of the affected areas. The lack of development of the upper and lower jaws can cause dental malocclusion, which requires surgical and/or orthodontic corrections. So it requires a multidisciplinary approach and long-term follow-up to monitor the growth and development.

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Conflict of Interest: Nil

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