

Goldenhar syndrome: A report of a rare case

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Abstract

Goldenhar syndrome is a rare condition described by Goldenhar in 1952. It is characterized by a combination of anomalies: dermal epibulbar cysts, auricular appendices, malformation of the ears, hypoplasia of malar bones, mandible and zygomatic arch. The etiology of this rare disease is not fully understood, as it has shown itself variable genetically and of unclear causes. In this report we present a case of Goldenhar syndrome in a 4-year-old boy, who presented all classical signs of this rare condition.

Key words; Goldenhar syndrome, Facial palsy, Anophthalmos, Microtia

Introduction

Goldenhar syndrome is a rare condition and the estimated incidence of this disorder is 1 per 5800 births with male: female ratio of 3:2. It is a presumably inherited condition, causing morphological abnormalities in the parts that developed from the first and second branchial arches during blastogenesis. It has a multifactorial etiopathology that includes nutritional and environmental factors¹.

Goldenhar first described this condition in 1952 as a disease that presents a combination of several anomalies such as dermal epibulbar tumors, peri-auricular appendices and malformation of the ears. It is also referred to as oculo-auriculovertebral (OAV) dysplasia and hemi facial microsomia².

The characteristic features of this syndrome are epibulbar dermoids, dacryocystitis, auricular abnormalities, preauricular appendages, preauricular fistulas, hypoplasia of the malar bones, mandible and zygomatic arch. Some associations recorded in the literature are macrostomia, micrognathia, high vaulted cleft palate, bifid tongue, malocclusion and other dental abnormalities³.

Some authors have also pointed out facial muscle hypoplasia, vertebral abnormalities, anomalies of the eyes¹, disturbances of the central nervous system, visceral anomalies⁴, Cardiac⁵ and genitourinary⁶ abnormalities.

Facial palsy⁷ though rare has been associated with it. A case of Goldenhar Syndrome, with association of anophthalmos and calcification of falx cerebri is also reported⁸.

The presence of anomalies of the ear (microtia) and of appendices on the ear is necessary for diagnosis of this syndrome.

In this article we present a case report of 4 years old boy diagnosed with Goldenhar syndrome.

Case report

A 4 years old boy reported to our clinics for routine dental treatment. The child was born to a young healthy parents (non-consanguineous). The prenatal and antenatal period was un-eventful and there was no relevant family history.

The physical features of the child was atypical with facial asymmetry, coloboma of middle 3rd of upper right eyelid, microtia with accessory preauricular tags, malar hypoplasia and severe unilateral mandibular hypoplasia (Fig 1, Fig 2 & Fig 3A). History revealed that right sided macrostomia was surgically closed and the scar was seen on the corner of the mouth (Fig 3B) and epibulbar dermoids had been surgically removed by Department of Ophthalmology when the patient was six months old. No behavioral problem was noted during examination.

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Fig 1: Right malar hypoplasia and asymmetry of face



Fig 2: Coloboma of middle 3rd of upper right eyelid



Fig 3: Right side of the face showing

- (A) Preauricular tags and deformed pinna
- (B) Scar resulting from surgical closure of macrostomia



Fig 4: Deviation of mandible to right side on opening the mouth

The patient was already evaluated by a Paediatrician and an Ophthalmologist. His cardiac function, eyes and sight were found to be normal. Audiological testing reports showed that both auricular structures and hearing were normal.

Dental examination revealed normal intra-oral soft tissues and all deciduous teeth were present. Few of the teeth in the left mandibular molar and upper anterior region were decayed. Mouth opening was not restricted, but the mandible deviated to the right side on maximum opening (Fig 4).

The patient was subjected to many examinations and treatments since birth and thus was very uncooperative at present in the clinical set up. Hence the patient was very uncooperative and the radiographic evaluation was not possible in this case.

Discussion

In approximately 10 to 33 percent of affected individuals, malformations are bilateral affecting both sides of the body, with one side typically more affected than the other (leading to asymmetry). In the majority of such cases, the right side is more severely affected than the left¹⁰. The presented case also showed right side of the face more severely affected than left side.

Goldenhar syndrome can be distinguished by varying degrees of vertebral, cardiac and sometimes structural kidney defects which were not present in this case. The patient exhibited clinical characteristics of complex Goldenhar syndrome with, including facial asymmetry, hypoplasia of the mandible, malar hypoplasia, macrostomia, preauricular tags, coloboma of eyelid.

Despite the reported frequency of cardiovascular alterations ranging from 5 to 58%, the reported patient had no cardiovascular alterations. Hearing disturbance or Facial nerve dysfunction were also not present.

Most cases have sporadic autosomal dominant and autosomal recessive modes of inheritance. Trisomy of 7, 22 have been described in association with Goldenhar syndrome¹¹. Reports indicate that several teratogenic agents such as retinoic acid, primidone, and thalidomide, have produced this syndrome in infants born to pregnant women exposed to those agents¹². In our case there was no history of maternal drug intake, any febrile illness or diabetes during pregnancy.

Although this syndrome presents some similarities with the Treacher-Collins syndrome, it is now considered a distinct entity. The features of Treacher-Collins syndrome are most frequently bilateral without ocular and aural anomalies¹³.

Delayed development of a tooth has been encountered in some cases although this is rare¹⁴ but was not observed in the present case. In the present case the deviation of the mandible to the right was due to the lack of the vertical and sagittal mandibular development, causing the mandible to rotate bodily to the right.

Prognosis of this disease is good in otherwise uncomplicated cases without any systemic associations. The treatment of the disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases.

The dental treatment rendered to this child was restoration of the decayed teeth. Maxillary and mandibular orthognathic surgery is indicated followed by future orthodontic treatment. Reconstruction of mandibular hypoplasia can be done with rib grafts and underdeveloped maxilla can be lengthened by bone distraction device in future. Plastic surgery for coloboma of eye and reconstruction of external ear has been planned to be done at 6-8 yrs of age by an ophthalmologist.

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