

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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