

Goldenhar's Syndrome (Oculo-Auriculo-Vertebral Dysplasia) with Congenital Facial Nerve Palsy

Nilufer Berker¹, Golge Acaroğlu², and Emel Soykan¹

¹Department of Uveal and Retinal Diseases, Social Insurance Eye Hospital, Ankara, Turkey; ²Department of Neuro-ophthalmology and Orbital Disease, Social Insurance Eye Hospital, Ankara, Turkey.

Goldenhar's Syndrome (oculo-auriculo-vertebral dysplasia) is a wide spectrum of congenital anomalies that involves structures arising from the first and second branchial arches. In this report, a case of a male infant, with the features of hemifacial microsomia, anotia, vertebral anomalies, congenital facial nerve palsy and lagophthalmos is described. Although the syndrome itself is not uncommon, the presence of congenital facial nerve palsy, which has been reported in rare cases, prompted this case report.

Key Words: Goldenhar's syndrome, oculo-auriculo-vertebral dysplasia, congenital facial nerve palsy, hemifacial microsomia, anotia

INTRODUCTION

Goldenhar's syndrome (Oculo-auriculo-vertebral dysplasia) consists of a constellation of anomalies, mainly affecting the first and second branchial arch derivatives in a sporadic manner.^{1,2} The known rate of incidence is approximately 1 in 5000 to 25,000 live births, and it is more common in males.³

Arlt first described the syndrome in 1845, but went largely unnoticed until 1952 when Goldenhar reported three new cases.³ Later, the range of clinical manifestations were extended, and a variety of terms have been used to describe the condition.⁴

Commonly observed malformations of Goldenhar's Syndrome include; auricular anomalies, such as preauricular tags, microtia, anotia, uni-

lateral maxillary and malar hypoplasia, epibulbar dermoids and vertebral anomalies.² In this case report, the case of an infant, with congenital facial nerve palsy and associated lagophthalmos, in addition to the other cardinal features of Goldenhar's Syndrome, are defined.

CASE REPORT

A 2-months old male infant was admitted to our clinic due to lagophthalmos in the left eye. He was born to a 36-year old woman, G1 P0, by cesarean section, performed after 38 weeks of gestation. The history revealed that the pregnancy had been complicated by gestational diabetes mellitus, which was controlled with a diabetic diet. Maternal fevers, ethanol, tobacco and radiation exposure were denied. The family history was noncontributory, and there was no parental consanguinity.

At birth, the infant weighed 2800 grams, which was appropriate for his gestational age. The Apgar scores were 7 and 9 at one and five minutes, respectively. He suffered no respiratory distress. A physical examination showed left anotia (Fig. 1), left facial asymmetry and the features of left facial nerve palsy, including lagophthalmos of the left eye, left naso-labial fold flattening and mouth corner drooping on the left side (Fig. 2). When crying, the mouth was drawn to the opposite (right) side, with the left half of the forehead remaining smooth (Fig. 3). The cerebral and orbital magnetic resonance images (MRI) were reported as normal. A temporal bone MRI was performed to define the etiology of the facial

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Reprint address: requests to Dr. Nilufer Berker, Huseyin Onat Sokak, 10/6, Asagi Ayranci, 06540, Ankara, Turkey. Tel: 90-312-4662870, Fax: 90-312-3126261, E-mail: niluferberker@hotmail.com



Fig. 1. Left anotia of the patient.



Fig. 3. The facial features of the patient while crying: The mouth is drawn to right, the left half of the forehead is smooth.



Fig. 2. Left lagophthalmos, hemi facial microsomia and nasolabial fold flattening of the patient while sleeping.



Fig. 4. Hemi vertebrae and scoliosis of the patient.

nerve palsy; however, the facial nerve components and facial canal were reported to be normal.

Further examinations revealed left choanal atresia, an asymmetrically localized uvula, left external auricular canal atresia, patent foramen ovale, pectus excavatus and vertebral anomalies, including hemi vertebrae at C7 - T6, and thoracic scoliosis deviated toward the right (Fig. 4). These

anomalies were consistent with a diagnosis of Goldenhar's syndrome accompanied by congenital peripheral facial nerve palsy. At 4 months of age, the patient suddenly experienced respiratory distress and high fever due to bronchopneumonia, and died one day later.

DISCUSSION

Goldenhar's Syndrome is a complex of con-

genital malformations, involving a broad variety of defects in structures of varying severity, derived from the first and second branchial arches. Poswillo⁵ hypothesized the pathophysiological mechanisms of these defects as a reduced blood supply, or focal hemorrhage, in the region of the developing branchial arches. Its occurrence is predominantly sporadic, in approximately 1 in 5000 to 25,000 live births,^{3,6} with reports of autosomal dominant transmission in 1 to 2% of cases.⁷ Several case series have reported on the teratogenic effects of gestational diabetes mellitus on the developing embryo, with an increased risk for Goldenhar's Syndrome in infants born to mothers with gestational diabetes mellitus.⁸⁻¹⁰ Presumably, this might have been a risk factor in our case.

The cardinal features of Goldenhar's Syndrome include ocular, auricular, vertebral and cranio-facial anomalies.^{1-3,8,11} Epibulbar dermoid and lipodermoid are the most common ocular anomalies; however, they did not occur in our case. Baum and Feingold¹² reported a 50% incidence of ocular anomalies. Lipodermoids are usually localized in the inferotemporal epibulbar area. These lesions may be missed on computed tomography (CT) and MRI since they are similar to orbital fat.

Auricular defects, including preauricular tags, microtia and anotia, are reported in 65% of cases.^{1,2,13} In our case, left anotia and external auditory canal atresia were observed with an accompanying left sided conductive hearing loss. Wang, et al. reported a 46% incidence of conductive hearing loss in their study.⁸

Vertebral anomalies, such as absence of vertebrae, hemi vertebrae, fused ribs, kyphosis and scoliosis, may occur in up to 60% of cases with Goldenhar's Syndrome.^{2,3,11} In this case, radiological investigations revealed hemi vertebrae at C7-T6, with right-sided scoliosis of the thoracic spine.

Craniofacial anomalies, including hemi facial microsomia, malar and maxillary hypoplasias, are found in 50% of patients with Goldenhar's syndrome. Facial nerve palsy is an important finding associated with craniofacial abnormalities,¹⁴ which have also been suggested to be a part, or a variant of Goldenhar's syndrome.¹⁵ Features, such as facial asymmetry, lagophthalmos, and naso-labial fold flattening, were associated with congenital

peripheral facial palsy in our case, was and was more obvious when the baby cried, as the mouth was drawn to the opposite side, and ipsilateral forehead remained smooth. Wang et al⁸ reported the incidence of facial nerve palsy in their patients with craniofacial abnormalities to be 33.3%. Carvalho and Song¹⁶ investigated the relation between facial nerve dysfunction and hemifacial microsomia, and reported a 22% incidence of facial nerve palsy. In most cases, the etiologic investigations of facial nerve palsy reveal an absence of physical or radiographic findings.^{14,17} The temporal bone MRI of our case showed normal components of the facial nerve and facial canal.

Facial palsy, associated with congenital malformations, has a poor prognosis for recovery.¹⁴ It affects the eye, causing lagophthalmos, ectropion, decreased tear production and corneal damage.¹⁸ In these cases, great attention should be paid to prevent corneal drying, especially at night. The first choice of treatment in our case was artificial tears and ointments, as the cornea of our patient was only minimally exposed while sleeping. Tarsorrhaphy is suggested for severe forms of lagophthalmos.¹⁸

The syndrome also affects other organs, especially the cardiovascular, genitourinary and pulmonary systems.² Cardiovascular system defects are the most common among these, with a reported rate of 36.7%.⁸ A patent foramen ovale was the only cardiovascular anomaly in our case. There were no pulmonary malformations. The cause of death was bronchopneumonia, at four months of age. In the previous case reports, most of the patients with Goldenhar's syndrome died in the first few months of their lives, and the common reasons of death was cardiopulmonary distress, due to cardiac and pulmonary malformations and bronchopneumonia.^{1,2,11}

This report describes a demonstrative case of Goldenhar's syndrome, mainly characterized by hemi facial microsomia, facial asymmetry, lagophthalmos, anotia and hemi vertebrae. Our aim was to draw attention to the association of gestational diabetes mellitus with craniofacial anomalies, and to emphasize that peripheral facial nerve palsy is also a part of this syndrome.

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