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Goldenhar complex associated with anal atresia: Brief case report

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Özet

Anal atrezi ile birlikte Goldenhar kompleksi olgusu

Burada anal atrezi ile birlikte "Goldenhar kompleksi" saptanan bir yenidoğan sunulmaktadır. Bu olgu "Goldenhar kompleksi"ne ek olarak doğumsal gastrointestinal anomaliler içermesi özelliğini taşımaktadır.

Anahtar kelimeler: *Goldenhar kompleksi, anal atrezi*

Summary

The authors report on a neonate who had anal atresia associated with Goldenhar complex. This report adds a further detail to the combination of this complex with additional congenital gastrointestinal anomalies.

Key words: *Goldenhar complex, anal atresia*

Introduction

The Goldenhar complex (oculo-auriculo-vertebral complex) was originally described as a nonrandom association of auricular, vertebral, and ocular defects [1]. The facial phenotype is characteristic and defined as hemifacial microsomia. Associated anomalies have been observed in cases with Goldenhar complex. Herein, we report on a neonate who had anal atresia and Goldenhar complex.

Case Report

A newborn baby, born after an uncomplicated twin pregnancy at 39 weeks of gestation with caserean section, was admitted to our department due to his facial anomalies and imperforated anus.

The baby weighted 2400 g at the time of admission. He had respiratory insufficiency and required mechanical ventilation. On examination, the patient was noted to have right facial asymmetry, absent (agnesia) right auricula and preauricular tags on the right side, closed eye fissure and anophtalmia on the right, microtia and hypoplasia of mandible on the right and anal atresia. He had also hexadactyly on the right hand (Figure 1).

Roentgenograms revealed various vertebral anomalies (Figure 2). There were blocked vertebrae at the levels of dorsal 4, dorsal 5, dorsal 12 and lumbar 1. At dorsal 8-9 a supernumerary vertebrae was found on the right, while hemivertebrae were seen at the level of dorsal 4 and 5. These anomalies caused thoraco-lumbar congenital kyphoscoliosis. Additionally there were absence of the right first rib as well as fusion and fork variation of the right 2nd and 3rd ribs. Right lung was hypoplastic. Other findings included, right orbital hypoplasia and ipsilateral hypoplasia of ramus mandible.

Routine laboratory values were normal. Invertogram revealed an intermediary type anal atresia and a right transverse colostomy was performed on an emergency basis. After a complicated postoperative course, needing mechanical ventilation and intensive care support the patient died at 21 days of age from respiratory and cardiac insufficiency. The family refused an autopsy.

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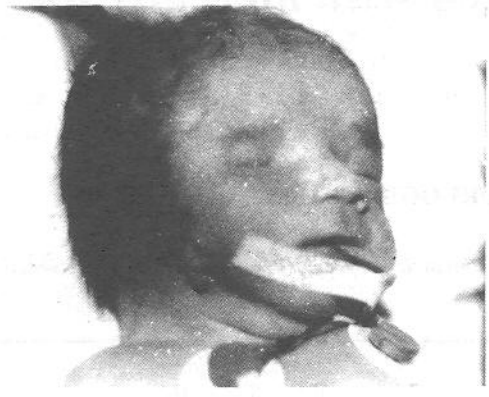

Figure 1. Facial asymmetry, preauricular tags and agnesia of the right auricula.

Discussion

We report on a case of Goldenhar complex (called as oculo-auriculo-vertebral complex) in association with anal atresia. Goldenhar complex is mainly characterized by extrafacial abnormalities, including CVS, cardiovascular system, and upper limbs and is a heterogenous condition [5]. Monozygotic twins with Goldenhar complex have been frequently reported [4]. Likewise our case had a healthy monozygotic twin brother.

This complex is quite variable and the patients could be affected by other anomalies, but the real incidence of extrafacial anomalies is not well known [6]. Although tracheoesophageal anomalies were seen in combination with Goldenhar complex [5], an association with anal atresia had been reported very rare in the literature [2,3].

Several theories such as, early vascular disruption and local hemorrhage resulting in destruction of differentiating tissues in the first and second branchial arch region or subsequently the theory, which describes this complex as a defect of blastogenesis, which took place during the first 4 weeks of gestation; have been suggested to explain the pathogenic mechanism of the anomalies observed in Goldenhar complex [7]. However these aspects are not addressed in this review. We think, that detailed investigations of cases similar to the one described herein might prove the value in establishing the etiology of this combination with this complex.

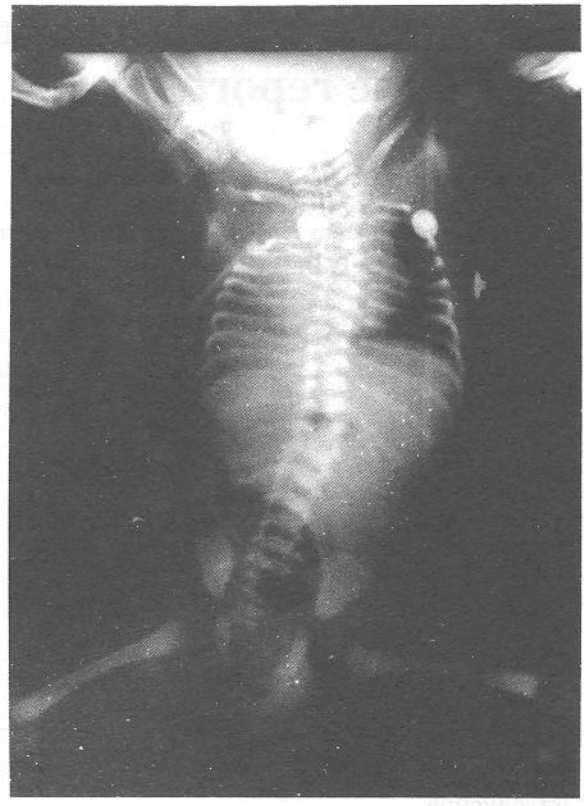

Figure 2. Direct X-ray examination of the baby.

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