European Review for Medical and Pharmacological Sciences 2012; 16: 1307-1308

Letter to the Editor

Goldenhar syndrome: a new case expanding the phenotype by costal agenesis and pulmonary hypoplasia

Dear Editor,

The proband, conceived through *in vitro* fertilisation (intracytoplasmic sperm injection), was born to non-consanguineous parents at 33 2/7 weeks of gestation by cesarean section. The pregnancy was a dichorionic diamniotic twin pregnancy and the case was the first-born twin.

The prenatal history revealed abnormal ultrasonographic (US) findings in one of the fetuses at 18th wk-of-gestation which were confirmed at 22nd and 28th wk-of-gestation including oligohydramnios, short vertebrae, kyphoscoliosis, right hydroureteronephrosis, left renal agenesis. There was no teratogenic insult. The parents denied antenatal diagnostic procedures and the babies were delivered. The first-born baby was admitted to the intensive care unit (ICU) due to prematurity, respiratory distress and major malformations. His birthweight was 1720 g (50th percentile), length was 37.5 cm (< 10th percentile) and head circumference was 30 cm (50th percentile). On physical examination, there were skin tags on the neck and right preauricular area (Figure 1), facial asymmetry, dysplastic right ear, cleft palate, short neck, single umblical artery and a bulging mass on the left side of the chest covered with intact skin (Figure 2). Chest X-ray showed near-complete costal agenesis of the left hemithorax except first and second ribs, an ipsilateral mediastinal shift, and pulmonary hypoplasia (Figure 3). Chromosomal analysis revealed a normal male karyotype. The infant died due to respiratory failure at 26 hours-of-age. The other twin was normal and was discharged home.

The baby fulfilled the diagnostic criteria of Goldenhar syndrome with facial asymmetry, dysplastic ear, skin tags, cleft palate, short neck. He had additional findings of left renal agenesis, costal agenesis and pulmonary hypoplasia.

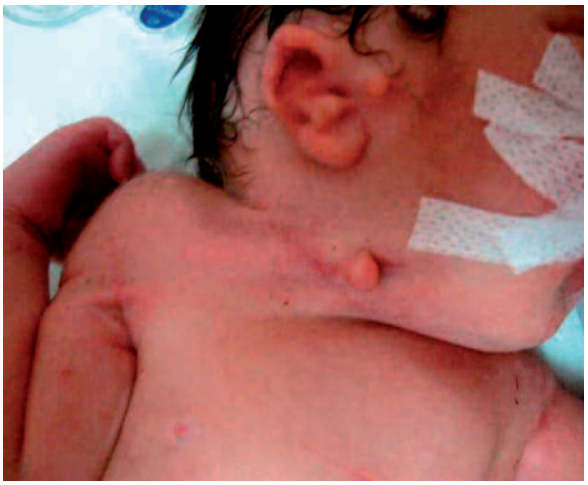

Figure 1. Skin tags at preauricular area and neck.

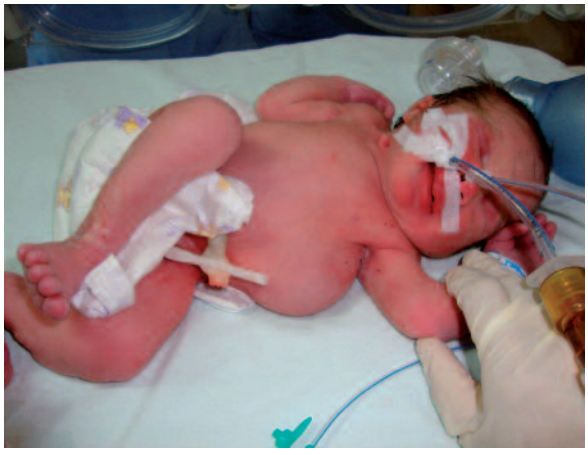

Figure 2. Protrusion of abdominal organs on the left side of the chest.

Corresponding Author: Ferhat Cekmez, MD; e-mail: ferhat_cocuk@hotmail.com

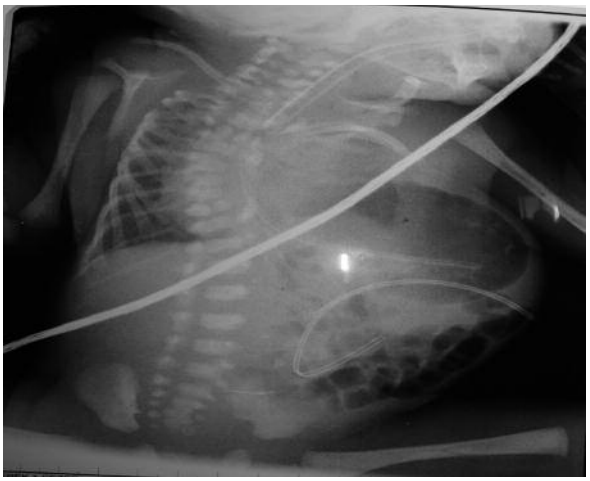

Figure 3. Complete costal agenesis of the left hemithorax, mediastinal shift and pulmonary hypoplasia.

Goldenhar syndrome, also known as oculo-auriculo-vertebral spectrum, is a complex, heterogeneous condition characterized by abnormal development of facial structures derived from the first and second branchial arches of the embryo. Associated anomalies also include asymmetry or hypoplasia of the face or mandible, unilateral epibulbar dermoids, colobomas of the upper lids, vertebral anomalies, and lateral facial clefts[1].

The association of pulmonary agenesis with facial microsomia has been described in a few cases and the syndrome with pulmonary agenesis is termed an expanded Goldenhar complex[2,3,4]. Here we report the first case of Goldenhar syndrome in the literature whith an additional finding of costal agenesis.

References

1) Gorlin RJ, Cohen MM, Levin LS. Branchial arch and oro-acral disorders. In: Syndromes of the head and neck. 4th ed. New York: Oxford University Press 2001; pp. 101-114.

2) Milani D, Selicorni A. Right pulmonary agenesis with ipsilateral microtia: a new laterality association or part of the oculoauriculovertebral spectrum? Prenat Diagn 2002; 22: 1053-1054.

3) Downing GJ, Kilbride H. An interesting case presentation: pulmonary malformations associated with oculoauriculovertebral dysplasia (Goldenhar anomalad). J Perinatol 1991; 11: 190-192.

4) Greenough A, Ahmed T, Broughton S. Unilateral pulmonary agenesis. J Perinat Med 2006; 34: 80-81.

*F. Cekmez, E.Z. Ince, A. Coban, S. Yildirim, E.Ö. Bulut, H. Kayserili**
Neonatology Department, Istanbul Medical Faculty, Istanbul, Turkey
*Genetic Department, Istanbul Medical Faculty, Istanbul, Turkey