Polymalformative syndrome with congenital heart defect

Síndrome polimalformativa com cardiopatia associada

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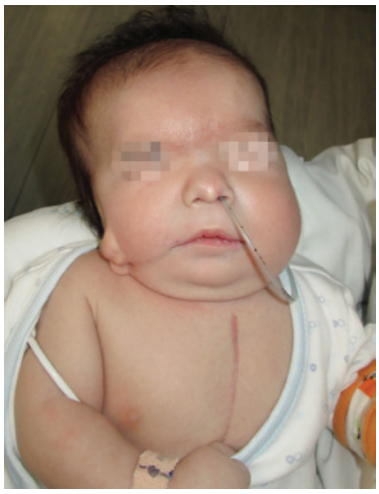

Figure 1. Hypoplasia of right-sided hemifacial, microtia, absence of zigomatic arcade, mandible and maxillary hypoplasia

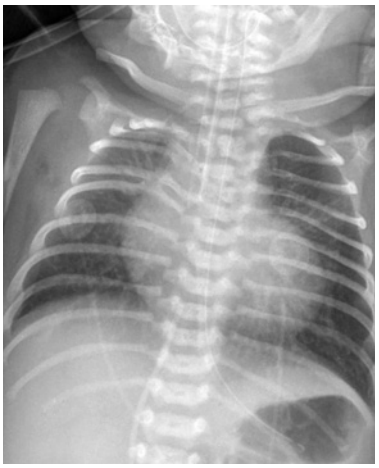

Figure 2. Radiographic image showing cardiomegaly, dorsal hemivertebra, fusions to level of first sixth right arches and apparent malformation of right mandible

This is the case of a male newborn whose mother during pregnancy was treated with chloropromazine, topiramate and diazepam, but without any other relevant family history, especially related with congenital malformation. At 34 weeks of gestation, the fetus was diagnosed with transposition of the great arteries (TGA). He born at 37 weeks of gestation, his birth weight was 3,229g and Apgar score 1/7/8. After delivery the he was maintained on invasive ventilation. The postnatal confirmation of TGA was carried out with restrictive foramen ovale throughout Rashkind septostomy done within the 2 hours after birth, under E1 prostaglandin therapy. At 13 days after birth, arterial switch operation was performed without significant intercurrences. At birth we observed hypoplasia of right-sided hemifacial, microtia, absence of zigomatic arcade, mandible and maxillary hypoplasia (Figure 1). Ophthalmology assessment highlighted slight ocular asymmetry. The chest radiography showed malformation of dorsal spine with hemivertebrae and fusion of ribs (Figure 2). His renal echography did not reveal malformations. Based on these findings the diagnosis was hemifacial microsomia. We conducted a pluridisciplinary follow-up for surveillance of complications, early intervention and schedule of correction of facial malformation.

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The hemifacial microssomia also known as oculo-auriculo-vertebral spectrum or Goldenhar syndrome is a result of a change in the development of first and second branchial arches, which is a possible consequence of vascular lesion at an early phase of pregnancy.[1,2] Most of situations are sporadic and family cases are also described in the literature.[3] This syndrome is characterized by combination of auricle malformation and hypoplasia of facial bones. A great phenotypic variability is show and it can be associated to anomalies of the spine, central nervous system, kidneys and heart.[1,2] Heart disease are described in 5-58% of the patients, being the most frequent the atrial and septal defects or ventricular and ventricular outflow tract.[4] With this case, we emphasize the prenatal diagnosis of a TGA that occurred in a sporadic situation of hemifacial microsomia.

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