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CLINICAL CASE

Spectrum of hemifacial microsomia in a pre-term newborn. Case presentation and literature review

A.Y. Medina-de la Cruz[a], I. Rodríguez-Balderrama[a,*], C.H. Burciaga-Flores[b], M.E. de la O-Cavazos[c]

[a] *Neonatology Service of the ''Dr. José Eleuterio González'' University Hospital, Autonomous University of Nuevo Leon, Mexico*
[b] *Genetics Service of the ''Dr. José Eleuterio González'' University Hospital, Autonomous University of Nuevo Leon, Mexico*
[c] *Pediatrics Department of the ''Dr. José Eleuterio González'' University Hospital, Autonomous University of Nuevo Leon, Mexico*

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KEYWORDS
Hemifacial microsomia;
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Abstract
Introduction: The spectrum of hemifacial microsomia, or facio-auriculo-vertebral spectrum, is a complex of craniofacial and vertebral anomalies. Axis malformation is microtia, more often on the right side of 3:2. It may be associated with mandibular hypoplasia and vertebral malformations. It is more frequent in males and in twin pregnancies.
Clinical case: Newborn male, preterm, of 29.5 weeks gestational age, twin product, second twin pregnancy, dichorionic and diamniotic, born by cesarean section, which presented hemifacial microsomia, microtia of the left Tanzer 3 and the right auricle of low implantation with a backward rotation, left appendices and macrostomia. A thoracoabdominal X-ray found hemivertebrae in the cervical and dorsal area, which discussed genetic performing diagnosis of the hemifacial microsomia spectrum. An ear TAC is done, the bone atresia of the left ear meeting at the level of the left ear without evidence of tympanic membranes and with a dysplastic oscicular chain attached to the lateral wall of the attic. Discharged at 78 days of chronological age with 6 days of age, corrected with the consultation of neonatal high-risk follow-up.
Conclusion: Facial asymmetry must be widely evaluated in patients with microtia, including deliberate search of renal, cardiac and spinal-level conditions, in order to diagnose pathologies such as the spectrum of hemifacial microsomia early.

* Corresponding author at: Servicio de Neonatología, Departamento de Pediatría, Hospital Universitario ''Dr. José Eleuterio González'', Universidad Autónoma de Nuevo León. Av. Madero y Gonzalitos s/n, Colonia Mitras Centro, C.P. 64460 Monterrey, NL, Mexico.
Tel.: +52 8183872460.
E-mail address: irb442000@yahoo.com.mx (I. Rodríguez-Balderrama).

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Introduction

The spectrum of hemifacial microsomia, or facio-auriculo-vertebral spectrum, is a complex of craniofacial and vertebral anomalies first described by Goldenhar in 1952.[1] Axis malformation is microtia, and it may even be the only manifestation. Nevertheless, it can be found linked to mandibular hypoplasia and vertebral malformations.[1] Microtia is a malformation characterized by the absence of some parts of the outer ear, and in some cases the entire ear. It can include the external hearing conduct and may be unilateral or bilateral.[1] The unilateral form occurs in between 79 and 93% of cases.[1]

It has an incidence of 1:500 to 1:3000 live births, is more frequent in males at a 3:2 ratio, and has a dominant autosomal inheritance in 1–2% and a sporadic inheritance in 98%.[1,2] It is the result of a defect in blastogenesis involving the first and the second pharyngeal arch, at approximately 30–45 days of gestation, with the formation of the first arch, which contributes to the formations of the structure of the face, both mandibular and maxillary portions, as well as the auricular pavilions.[1–3] From the dysmorphologic point of view it is classified as a disruption. Among prenatal risk factors, there are: multiple pregnancies, anemia, advanced maternal age, threatened miscarriage, diabetes mellitus type 1 and 2 and medications such as isotretinoin. Twin pregnancy increases the risk of congenital malformations 2.47 times.[4]

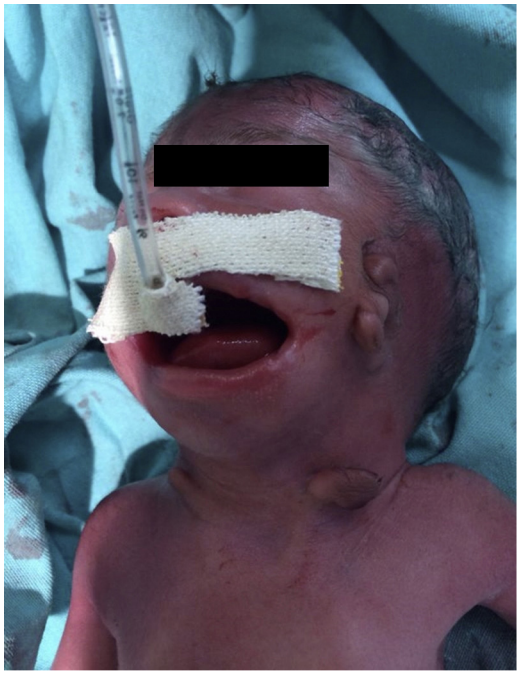
Among the milder cases, preauricular appendices or isolated microtia may occur, whereas in the most severe cases, macrostomia and epibulbar dermoid, also known as the Goldenhar syndrome, occur.[1,2] Hemifacial microsomia appears with a frequency of 20–65%. Regarding microtia, the right side is the most frequently affected, with a 3:2 ratio. Auricular malformations have a frequency of 65–99%, including preauricular appendices (with 40%).[1,2,4,5]

Axial skeleton alteration is limited to the cervical region and occasionally to the thoracic vertebrae, including hemivertebrae in 30%, renal malformations in 1–10% and cardiac malformations in 14–47%.[6,7]

There are known malformations associated with the spectrum, such as cleft palate, tracheoesophageal fistula, finger and hand anomalies and pulmonary hypoplasia.[8–10]

Case presentation

A male, newborn, preterm of 29.5 weeks of gestational age, twin product, twin number 2, the mother 23 and the father 24 years old, non-blood relatives, without exposure to teratogens, product of a third gestation. It was a dichorionic and diamniotic spontaneous twin pregnancy, with proper prenatal care, and went through the first trimester with threatened miscarriage. There is a premature rupture of membranes at week 29.4 of gestational age, a dosage of pulmonary maturation inductors and the patient is granted conservative management after not presenting preterm labor. However, 7 h after rupture there is fetal distress caused by type 2 decelerations of twin number 2, thus interrupting the pregnancy via C-section. The product is obtained with an Apgar score 5/8, a weight of 1110 g, size 39 cm, and a 27-cm head circumference, requiring admittance to the Neonatal Intensive Care Unit. Twin number 1, male born with



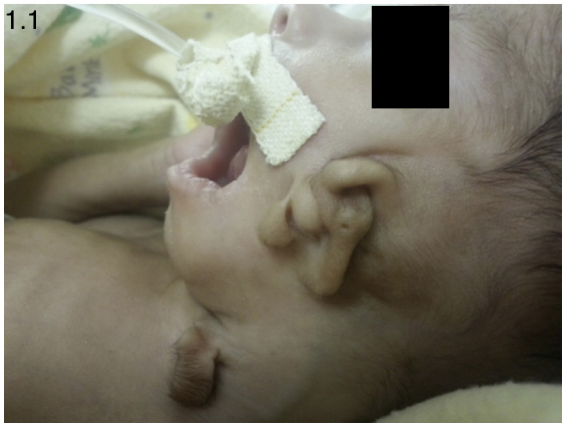


Figure 1 Photograph of the front. (1.1) Profile photograph.

a weight of 1290 g, size 39 cm, a 27.5 cm head circumference and an Apgar score of 8/9, had no dysmorphias detected during physical examination and no bone defects in imaging studies, thus, no genetic studies are performed. He dies after 5 days of life outside the womb due to prematurity-related complications; hyaline membrane disease grade 2, pulmonary hemorrhage and late sepsis, we requested an autopsy from the parents; however, they declined.

During twin no. 2's physical examination, a wide anterior fontanelle is detected, broad nasal bridge, hemifacial microsomia, anteverted nares, left microtia Tanzer 3, low implantation of right auricular pavilion with backwards rotation, bilobed left preauricular appendices, bilateral *fifth finger clinodactyly, bilateral cryptorchid, macrostomia, pointed palate, micrognathia without evidence of epibulbar dermoid* (Fig. 1).

A thoracoabdominal X-ray is taken, observing hemivertebrae in cervical and dorsal areas, in addition to hyaline membrane disease grade 2 (Fig. 2) requiring 3 doses of pulmonary surfactant and mechanical ventilation, accomplishing continuous positive nasal airway pressure and