

Epignato, sindrome del cuore sinistro ipoplasico e trisomia 18 in una gemella piccola per età gestazionale

Epignathus, hypoplastic left heart syndrome, and trisomy 18 in a small for gestational age female twin

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Riassunto

Descriviamo una rara associazione di sindrome del cuore sinistro ipoplasico, trisomia 18 ed epignato in una gemella di 970 g, nata a 37 settimane di età gestazionale. La bimba è deceduta dopo 24 ore di vita. I neonatologi ed i ginecologi dovrebbero prestare attenzione a questa rara associazione al fine di fornire un accurato counselling prenatale.

Abstract

We describe a rare association of hypoplastic left heart syndrome, trisomy 18, and epignathus in a 970 g female twin born at 37 weeks of gestational age. She died at 24 hours of age. Neonatologists and obstetricians should be aware of this rare association for a thorough prenatal counselling.

Introduction

Epignathus is an extremely rare form of neonatal teratoma which originates from the oral cavity and/or the pharynx. Very few cases are reported in the literature according the synonyms of oropharyngeal, nasopharyngeal, or facial teratomas. Most commonly it

originates from the palate and protrudes outside the mouth.¹ Rarely, epignathuses with also intracranial extension have been reported as well.²

Although epignathus is rarely malignant, its mortality is very high because of the local mass effect, chiefly airway obstruction (i.e. respiratory failure), and the severe associated anomalies which include facial clefts, bronchial cysts, congenital heart defects (double outlet right ventricle, univentricular heart, patent ductus arteriosus, atrial septal defect), trisomy 13, intracranial abnormalities, diaphragmatic hernia, renal and vertebral disorders.^{1,3} Since its first description in 1960, no association of trisomy 18 and epignathus have been reported so far.⁴

In this manuscript we present a rare case of epignathus associated with hypoplastic left heart syndrome (HLHS) and trisomy 18, in a 970 g twin term female infant.

Case Report

This was the second, female twin of a bicorial/biamniotic pregnancy, conceived with *in vitro* fertilization, and born to a nonconsanguineous Caucasian couple with no significant family history. At 24-weeks gestational age, a fetal ultrasound showed an oral mass (1.4 x 1.4 cm) in one twin, also affected with HLHS, and severe intrauterine growth retardation. The other twin was a healthy female fetus. In the following weeks, the oral mass increased its size, polyhydramnios developed, and a diagnosis of a potential sublingual lymphangioma was made. Ethical issues were discussed with parents, who did not release consent for amniocentesis, and an elective cesarean section was planned at 37 weeks of gestational age. At birth, the affected twin showed a giant mass protruding from the oral cavity and, covered by skin, from the right side of neck. The mass protruding from the mouth had a regular surface covered by arteries, veins, and the tongue surface on top of it (Fig. 1). The patient was intubated through the mouth with a laryngoscope, she was connected to mechanical ventilation and received treatment with prostaglandins. APGAR scores were 4 and 6 at 1 and 5 min-

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Figure 1.

The epignathus protruding from the oral cavity of the infant, the endotracheal tube, the umbilical arterial and venous catheters.

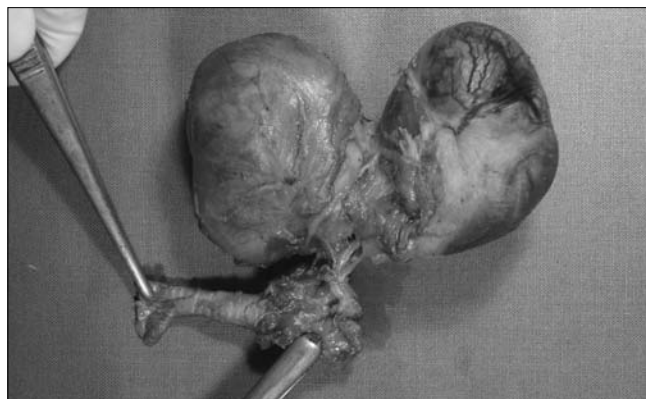


Figure 2.

Autoptic appearance of the epignathus.

utes, respectively, and birth weight was 970 g. The external neonatal deformities included caudal displacement of the mandible, which could be visualized on the X-ray of the mass, clinodactyly of the 5th finger of the hands, transverse palmar creases, lower set ears, left pre-auricular tags, and the general appearance of a small for gestational age infant. Standard karyotype showed 47 XX, +18. Head ultrasound was normal and cardiac ultrasound confirmed HLHS with patent ductus arteriosus, and arterial septal defect. Soon after birth, progressive anuria, and metabolic acidosis developed, until she died at 24 hours of age. No attempts of surgical excision of the mass or palliative correction of the heart defect were done, because of her low birth weight and unstable condition.

The autopsy confirmed hypoplastic left ventricle, hypoplastic ascending aorta, and aortic arch, mitral and aortic valve stenosis, dilatation of right atrium and ventricle. Trachea, main bronchi, and lungs were normal. Postmortem total excision of the mass was possible: it appeared to originate from the oral pharynx, at the root of the tongue: one lobe (4 x 4.5 cm) covered by the external surface of the tongue protruding out of the mouth, and one lobe (4 x 5 cm) covered by the skin and protruding under the mandible through the right side of the neck (Fig 2). The histology showed the following tissues: immature neural-epithelial tissue, mature glandular tissue with mucipar columnar epithelium, and chondroid tissues. No mature organs were visible. All these findings were consistent with a pathological diagnosis of immature teratoma.

Her sister was healthy, with a birth weight of 3800 g, APGAR scores of 8 and 9 at 1 and 5 minutes, respectively and a standard karyotype of 46 XX.

Discussion

We have described a rare association of epignathus with HLHS and trisomy 18, in a small for gestational age term twin infant. The incidence of the epignathus tumor is between 1:35.000 and 1:200.000 live births; it accounts for 2-9% of all neonatal teratomas which occur approximately in 1:4000 live births.⁵ This tumor is composed

of tissues derived from ectodermal, mesodermal or endodermal layers, it is composed of solid or solid and cystic structures, and its histology might be mature or immature.⁵

Associated anomalies have been reported in approximately 6% of the cases⁶ and include facial clefts, intracranial abnormalities, diaphragmatic hernia, renal and vertebral anomalies.^{7,8} Polyhydramnios is usually present, secondary to the obstruction of fetal mouth and the inability to swallow. Among associated congenital heart defects HLHS has never been reported. Previously, it has been associated with univentricular heart, patent ductus arteriosus and atrial septal defect.³ Oral teratomas have been associated to trisomy 13⁹ but never to trisomy 18.⁴

The prognosis of this tumor depends mainly on the size of the tumor, the degree of airway obstruction, and the associated anomalies. Its histology is often benign, however fetal and neonatal death is very common due to the local mass effect, as it occurs also in the rare intracranial lesions or in the nasopharyngeal lesions.^{2,3}

Therefore, in case of a potential fetal epignathus, prenatal ultrasound and fetal magnetic resonance imaging (MRI) are needed to understand the lesion, its size, its relationship with near vital structures, associated hominous anomalies, and the route of delivery (cesarean section *versus* vaginal delivery). MRI is also needed to rule out other important differential diagnoses (i.e. encephalocele, meningoencephalocele, dermoid cysts, choriocarcinoma, epulis, yolk sac tumor, rhabdomyosarcoma, glioma, haemangioma).^{5,10} Amniocentesis should also be performed to rule out anomalous karyotypes which might worsen prognosis. At birth, a multidisciplinary approach is also often needed: a neonatologist to provide endotracheal intubation, and a pediatric surgeon to perform an urgent tracheotomy if intubation fails, during an EXIT procedure.

Conclusion

Neonatologists and obstetricians should be aware of the rare association we have described for a thorough prenatal counselling.

When an epignathus is diagnosed early in the course of pregnancy, the option to terminate the pregnancy should also be discussed with parents.

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