OEIS complex and diaphragmatic agenesis: a rare association

María Recuero Pradillo1*, Beatriz Moreno Torres2, Carlos Quimbayo-Arcila3, Esperanza Carabias Lopez1, Laura Sánchez Sánchez2, Ángel Pantoja2, Pedro Rafael Altungy Labrador3, Juan Ruiz Martín1
1Department of Pathology. Hospital Virgen de la Salud, Toledo, Spain
2Department of Pediatrics and Neonatology. Hospital Virgen de la Salud, Toledo, Spain
3Department of Research. Complutense University of Madrid, Spain.

Article Info

Article notes
Received: October 24, 2019
Accepted: March 31, 2020

*Correspondence: Maria Recuero Pradillo, Department of Pathology, Hospital Virgen de la Salud, 30 Barber Avenue, Toledo, Spain. 45004; E-mail: recuero_maria@hotmail.com.

©2019 Recuero M. This article is distributed under the terms of the Creative Commons Attribution 4.0 International License.

Keywords
omphalocele
meningomyelocele
imperforate anus
congenital malformations
twin pregnancy
diaphragmatic agenesis
congenital contractures

Introduction

OEIS complex, firstly described by Carey in 19781, is a severe anomalies association which includes: omphalocele, cloacal extrophy, imperforate anus and spinal defects. It constitutes a very odd disorder with a frequency of 1 case per 200,000 – 400,000 pregnancies, even lower frequency in twin’s pregnancies.

Material and Methods

An autopsy of a newborn was reviewed for the study. A detailed necropsy including external and internal postmortem examination was conducted. A review of both the mother’s gynaecological and neonatal paediatric clinical history, along with a bibliographic revision were performed too.

Case Report

40 years-old mother, first pregnancy, bi-chorionic and bi-amniotic twin pregnancy. In the 15-weeks prenatal echography, poliformations were detected in one of the fetus. The birth took place in the 36th week, through a caesarean performed due to buttocks presentation in both twins. First twin, male, was born with 2,500 gr. and good evolution. Second twin, polimalformed, was born depressed, Apgar 2/3, passing away fifteen minutes later. The baby weighed 1,600 gr. A clinical autopsy of the second twin is requested.

Prenatal cytogenic study: regular karyotype 46 XY.

In the external analysis there were evidences of no anus, lack of external genitalia– only vestiges as soft appendices –a lumbosacrus myelomeningocele with 11cm in diameter, giant omphalocele broken which contains liver and intestinal handles. The extremity exam revealed left upper hipoplasia, with flexion contractures of elbow, and bowing of the left wrist and ectrodactyly. Lower limb presented left-equinovarus foot.

In the internal exam: pulmonary hypoplasia, agenesis of the diaphragm with intrathoracic stomach, heart in a marked right deviation, left renal agenesis, absence of urorectal-septum, intra-abdominal gonads with aplastic testicular appearance, accessorial spleen.

Discussion

The fetal OEIS complex was combined with multiple malformations including cardiac defects, in our case right-heart was observed2.
Most patients with OEIS complex have associated lower extremity defect, apart from these, one very interesting finding in our case was the left superior member arthrogryposis. The exact aetiology of these congenital contractures is uncertain, although arthrogryposis is thought to be related to any factor that curtails fetal movement in utero therefore is noted an increased prevalence in twins\(^8\).

The differential diagnoses of disorders that consistent with omphalocele included OEIS complex and Pentalogy of Cantrell. The Pentalogy of Cantrell, is typically composed of pericardial or sternal defect and the absence of spinal defects allow differentiation from OEIS complex\(^2\).
Moreover, it was actually exceptional is the coexistence of OEIS Complex and diaphragmatic agenesis. In our literature review, we did not find any report of an association between both entities.

**Conclusions**

Current study presented the rare finding of coexisting OEIS complex and agenesis of the diaphragm in twin's pregnancy.

OEIS complex is an entity more frequent in males, being the cases usually sporadic. Some isolated cases in brothers, twin-pregnancy and in chromosomal-related alterations (trisomy in 18, 47, XXX; no-balanced translocation between 9q and Yq) have been described.

Prenatal diagnostic is possible when there is a defect in the abdominal wall, myelomeningocele and absence of bladder visualisation. Other secondary findings that may drive to prenatal diagnostic: kyphoscoliosis, equinovarus-foot, genitourinary alterations, single umbilical artery and bladder/spinal defects.

In physical examination, the case reported here gathered all caudal defects compatible with OEIS Complex. The coexistence with anomalies of upper body like diaphragmatic agenesis, arthrogryposis left arm and right-heart, which cannot be explained by the mechanism typically associated with OEIS Complex, thats why this case is exceptional.

In almost every case, prognostic is always fatal, depending on the structural defects severity. An early prenatal diagnostic allows it assessment by a multidisciplinary team, which may allow a more effective genetic advice and management.

**Conflict of interest**

The authors declare that they have no conflict of interest.

**Informed consent**

Informed consent was obtained.

**References**