

Case Report

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OEIS complex and diaphragmatic agenesis: a rare association

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Keywords

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twin pregnancy
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Introduction

OEIS complex, firstly described by Carey in 1978¹, is a severe anomalies association which includes: omphalocele, cloacal exstrophy, 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.

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40 years-old mother, first pregnancy, bi-chorionic and bi-amniotic twin pregnancy. In the 15-weeks prenatal echography, poli-formations 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, poli-malformed, 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 genitalias– 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 agnesis, absence of urorectal-septum, intra-abdominal gonads with aplastic testicular appearance, accessory 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 twins⁸.

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 complex³.



Figure 1: Posterior view of an infant with OEIS complex. Soft tissue mass in the right and lumbosacral lower back/buttock which is covered with skin and represents the myelomeningocele and left omphalocele. Left-equinovarus foot.



Figure 2: Posterior view of an infant with OEIS complex. Soft tissue mass in the right and lumbosacral lower back/buttock which is covered with skin and represents the myelomeningocele and left omphalocele. Left-equinovarus foot.



Figure 3: Anterior view of an infant with OEIS complex.



Figure 4: Ambiguous genitalia and lower abdominal wall defect containing abdominal viscera consistent with an omphalocele.



Figure 5: Arthrogryposis involving the left upper limb.

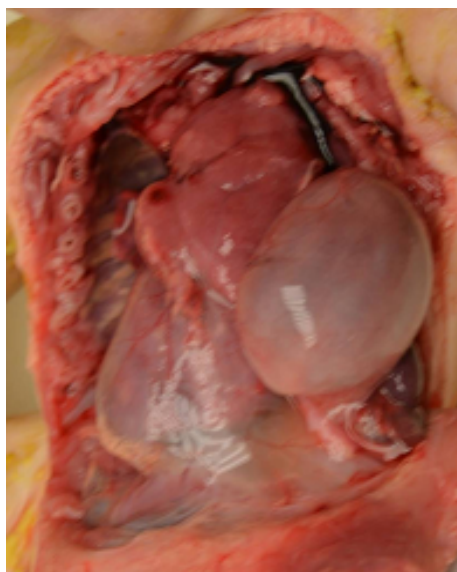


Figure 4: Right-heart.

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 chromosomic-related alterations (trisomy in 18, 47, XXX; no-balanced translocation between 9q and Yq) have been described⁴.

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 alterations⁵, single umbilical artery and bladder / spinal defects⁶.

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, that's why this case is exceptional⁹.

In almost every case, prognostic is always fatal, depending on the structural defects severity⁷. An early prenatal diagnostic allows its 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.

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