Case Report

OEIS complex—using MRI in diagnostic: Two case reports✩,✩✩

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A B S T R A C T

OEIS complex is an acronym for a specific, extremely rare, grouping of more commonly occurring congenital malformations consisting of an omphalocele (O), exstrophy of the cloaca (E), imperforate anus (I) and spinal defects (S). It is a midline defect occurring in early blastogenesis or in mesodermal migration. The incidence is not known, it has been estimated to be 1 in 200,000-400,000 pregnancies. The etiology is thought to be heterogeneous. Most cases occur sporadically but several reports show occurrence in siblings, concurrent occurrence in monozygotic twins, and more frequent occurrence in IVF pregnancies. We report two cases of OEIS complex. Case I was recognized postnatally in twin gestation. Case II, an IVF pregnancy, was diagnosed prenatally at 35 weeks gestation by ultrasound (US) and further confirmed by magnetic resonance imaging (MRI). The purpose of this report is to present two additional cases of this rare malformation, give further evidence of the OEIS complex occurring more commonly in multiple gestations (monozygotic twins) and IVF pregnancies and illustrate the importance of MRI in prenatal diagnostics in addition to US providing better prenatal counseling, perinatal care, and planning of reconstructive surgical management.

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Introduction

The OEIS complex refers to a group of congenital anomalies consisting of omphalocele, exstrophy of the cloaca, imperforate anus, and spinal defects. It is an extremely rare midline defect in early blastogenesis or mesodermal migration. The estimated frequency of the OEIS complex is 1 in 200,000-400,000 live births. Most cases develop sporadically, but a large number of studies report occurrence in siblings, simultaneous occurrence in monozygotic twins, and it also occurs more often in IVF pregnancies. The etiology of OEIS complex is suggested to be heterogeneous which means that both genetic and environmental causes are involved in its development. It is most frequently diagnosed in second-trimester screening using ultrasound.

In both cases, mothers denied being exposed to known teratogens or medications. Both parents were healthy, nonconsanguineous white couples with no family history of hereditary diseases and malformations.

Case 1: Unrecognized OEIS complex in twin pregnancy

A 32-year-old gravida 2 para 3, whose prior pregnancies were uncomplicated, was referred to our clinic at 16 weeks gestation. The first prenatal US at 14 weeks showed a monochorionic, monoamniotic twin gestation with the demise of one fetus. US exam at 16 weeks in our institution showed an anterior abdominal wall cystic formation (33 mm in diameter) near the umbilical cord insertion in the surviving twin (Fig. 1). Amniocentesis established a normal female (46XX) karyotype and the pregnancy was continued. At 35 weeks’ cystic structure was diagnosed as an omphalocele. Delivery was unremarkable at 39 weeks following a spontaneous vaginal birth of a vigorous female baby with birth weight and length of 3450 g (50th percentile) and 48 cm (25th percentile) respectively. Immediately after birth, the child was admitted to the neonatal intensive care unit (NICU).

In addition to the omphalocele, cloacal extrophy, imperforate anus, split clitoris with vaginal halves, rudimentary uterine horns, fallopian tubes, and ovaries located on each side were diagnosed postnatally. US demonstrated a horseshoe kidney and right hydronephrosis. X-ray revealed hypoplastic fourth cervical vertebrae and thoracolumbar vertebral malformations.

Case 2: Recognized OEIS complex in an IVF pregnancy

A 30-year-old primigravida, IVF fertilization, with good prenatal care and routine clinical and US examinations was referred to our institution at 35 weeks gestation with a suspected neural tube defect. US in our institution showed a lumbosacral meningomyelocele. The fetal bladder and vessels could not be visualized. A solid tubular anterior abdominal-wall mass was detected in close proximity to a single umbilical artery (US) cord. Fetal US suggested prenatal diagnosis of OEIS complex (Fig. 2). Further investigation included magnetic resonance imaging (MRI) using MR “Siemens, Eshree 1.5 T” machine.

MRI confirmed a lumbosacral neural tube defect 3 × 5 cm in size, omphalocele 6 cm in length (with liver and small bowel herniating into the sac), and absence of fetal bladder (Fig. 3). Brain ventricular system was mildly dilated at the ventricle trigonum bilaterally. Internal organs and limbs were normal. Defects in pelvic bones could not be excluded. Genital organs could not be clearly identified.

A vigorous baby with ambiguous genitalia was born by a scheduled c-section at 38 weeks, birth weight and length of 3149 g (50th percentile) and 49 cm (50th percentile) respectively. Immediately after birth, the child was admitted to the NICU of a tertiary care center for surgical correction.

In addition to the omphalocele and meningomyelocele, cloacal extrophy, protruding bowel, imperforate anus, atresia of the ascending colon, and a widened pubic arch were diagnosed postnatally. Karyotype was normal male (46XY). Explorative surgery revealed intraabdominal testes, glans penis, and bifid cavernous bodies.
However, early formation of twin pregnancies [1–3] has been related to chromosomal abnormalities, single gene defects, sporadic causes, and genetic etiology. The etiology of OEIS complex is unclear. It is believed to be caused by genetic and environmental factors. Most cases are sporadic and karyotype normal, as in our case. Familial cases, chromosomal abnormalities, and single gene defects have been reported [1–3] but the evidence is still lacking to suggest genetic etiology. Our first case shows a monozygotic twin gestation with vanishing twin syndrome and OEIS complex in the surviving twin. El Hattab et al. found 26 OEIS cases in twin pregnancies (18 monozygotic, 2 dizygotic, and 5 unknown zygosityes) with 50% concordance rate in monozygotic twins [2]. The concordance may support the theory that early malformation complexes are caused by the same disturbance in early blastogenesis [4] or a possible genetic contribution [4]. However, Nance suggested 2 explanations for discordance. A concordant twin may be aborted early in pregnancy which can conceal the twinning association of OEIS as could have been in our case or, the twinning process with its opportunities for asymmetry, cytoplasmic deficiency, and competition may favor discordant expression [5].

Case II associates OEIS with IVF fertilization. A recent report suggests the risk of major malformations in IVF is higher [6]. The role of IVF in epigenetic alterations or manipulation of the embryo ex utero might alter biochemical surroundings affecting embryogenesis [7,8] causing malformations such as OEIS.

Prenatal diagnosis of OEIS may be difficult. The full extent of abnormalities may not be clear until after birth. Our first case was not diagnosed prenatally. The majority of cases are diagnosed postnatally or during fetal autopsy. In the great majority of prenatally diagnosed cases, the diagnosis was made in the second trimester [9]. Differential US diagnosis with other midline abdominal defects may be challenging and includes pentalogy of Cantrell and limb-body wall com-
plex [10] which may represent a spectrum of developmental defects since there is a significant clinical overlap. Other differential diagnoses include amniotic band sequence and body stalk anomaly [11]. Ultrasound is usually the first imaging method used when prenatal suspicion of OEIS complex is made. Specific findings include a ventral wall defect, spinal defect and failure to visualize the fetal bladder [12]. The most specific finding is failure to visualize the bladder which differentiates OEIS from other abdominal wall defects. As mentioned above differential diagnosis with other midline abdominal defects can be hard. MR imaging is an excellent tool for providing a more exact diagnosis and a useful adjunct to US for complex fetal anomalies such as OEIS complex. MRI helps visualize fetal anatomy and structural defects more exactly. In our first case, we believe that if MRI had been used, prenatal diagnosis of OEIS complex would have been made. In our second case where suspicion was made with US, we were able to see more exactly the size and contents of the omphalocele (liver and intestines) and the extent of neural elements involved in the spinal defect. Using MRI in prenatal diagnostics we were able to confirm the diagnosis, provide better prenatal counseling, alert a tertiary center and plan reconstructive surgery in advance. Therefore, we recommend the use of MRI in all cases suspicious of OEIS complex where immediate invasive treatment is inevitable.

### Patient consent

The authors confirm that written, informed consent for publication of their case was obtained from the patients.

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