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A Rare of OEIS Variant Manifesting as Omphalocele, Anal Atresia, Scrotum Bifida, Cryptorchidism, and Hypospadias: A Case Report

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ABSTRACT

Omphalocele, exstrophy, imperforate anus, and spinal defects (OEIS) complex is a rare disorder results from abnormal midline development during early gestation and has a multifactorial etiology. We had a term male neonate with OEIS complex born to a 34-year-old G4P1A1 mother with a spontaneous conception and no notable medical or teratogenic history. Patient attended antenatal care four times with an obstetrician, and prenatal ultrasounds revealed an anterior abdominal wall defect suggestive of omphalocele. The patient underwent vaginal delivery at 37 weeks, resulting in a live male infant weighing 3200 g with Apgar scores of 4 and 8. Postnatal examination revealed a large omphalocele, absent anal opening, underdeveloped phallus, widely spaced scrotum with non-palpable testes, and no limb anomalies. Karyotyping showed a normal male chromosomal pattern. Surgical intervention, including exploratory laparotomy and colostomy, was performed on day 3 of life. Despite management, the neonate died at one month of age. OEIS complex has poor prognosis. Early prenatal diagnosis remains challenging but crucial. Multidisciplinary management, timely surgical intervention, and increased access to genetic counseling and antenatal care are essential to improve outcomes and support affected families.

Keywords: congenital anomalies, OEIS complex, prenatal diagnosis

INTRODUCTION

The OEIS complex is an uncommon congenital disorder that involves a combination of abnormalities, including omphalocele, cloacal exstrophy, imperforate anus, and spinal malformations.[1] Its occurrence is estimated at approximately one in every 200,000 to 400,000 live births, with a slightly higher frequency observed in males. The etiology is multifactorial, involving both genetic and environmental factors.[2] The condition results from abnormal ventral body wall closure and disrupted midline development between the 4th and 6th weeks of gestation. Key features include omphalocele, caused by the herniation of abdominal contents through the umbilicus; cloacal exstrophy, leading to malformations of the bladder, gastrointestinal tract, and genitalia; and imperforate anus, characterized by the absence of a normal anal opening.[3,4] Here, we present a case of an OEIS variant manifesting as omphalocele, anal atresia, scrotal bifida, cryptorchidism, and hypospadias.

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CASE REPORT

A 34-year-old woman, G4P1A1, with a non-consanguineous marriage and spontaneous conception, presented to our hospital in labor. She had attended antenatal care with an obstetrician four times, and antenatal ultrasounds were performed. There was no reported history of diabetes, other medical conditions, use of teratogenic medications, or any familial occurrence of congenital anomalies. She did not smoke or consume alcohol. On examination, the uterus measured 30 cm, with contractions occurring 2 times in 10 minutes, lasting 10 seconds each. The fetal heart rate was 128 bpm, and the fetus was in a head presentation. Vaginal examination revealed a cervical dilation of 3 cm, with the fetal head at -2 station and an adequate pelvis. Hematological and biochemical investigations showed values within normal ranges. An ultrasound examination has been performed. Ultrasound examination showed a single live intrauterine fetus corresponding to 37 weeks and 1 day of gestation, in cephalic presentation, with a protruding mass arising from the lower abdominal wall covered by a membrane and measuring 4.76

× 4.91 cm. The prognosis was discussed with the patient's family, who chose to proceed with vaginal delivery. A live male infant was delivered vaginally. He weighted BW 3200 g, BL 49 cm, A/S: 4/8 with multiple anomalies was seen.

On external examination: A term infant, corresponding to a gestational age of 40–42 weeks, with a birth weight of 3200 g and length of 49 cm. A large omphalocele was noted. The scrotal sac appeared well developed for the gestational age but was widely separated; however, the testes were not palpable within the scrotal sacs. The phallus was hypoplastic, displaying a distinct separation between the prepuce and glans. No anal opening was identified in the perineal region. No deformities of the limbs or joints were noted.

A babygram examination was performed, revealing distension and dilatation of the rectosigmoid. Karyotype analysis was conducted, and the metaphases counted and analyzed showed no structural or numerical chromosomal abnormalities. The karyotype was consistent with a male sex. He had surgery on the 3rd day, during which an exploratory laparotomy and colostomy were performed. Unfortunately, the baby passed away at the age of 1 month.

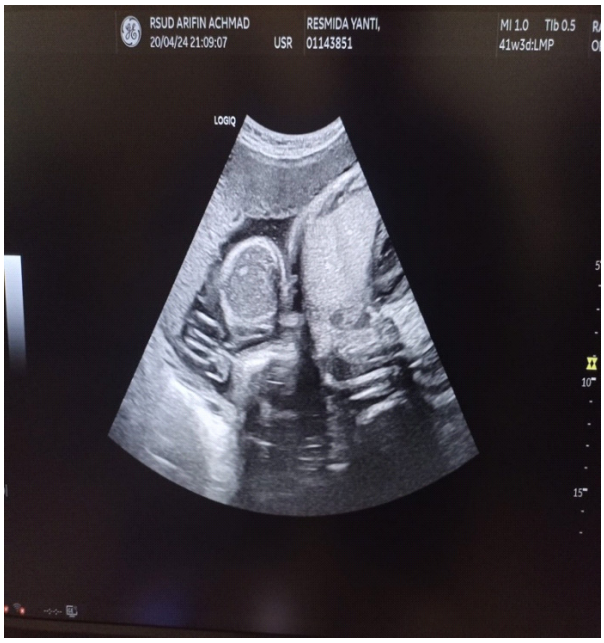

Figure 1. Pre-natal US examination showing a protruding mass is observed from the lower abdominal wall, covered by a membrane

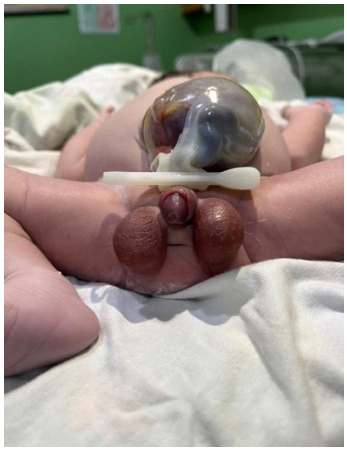

Figure 2. Anal Atresia, Scrotum Bifida, Cryptorchidism, and Hypospadias

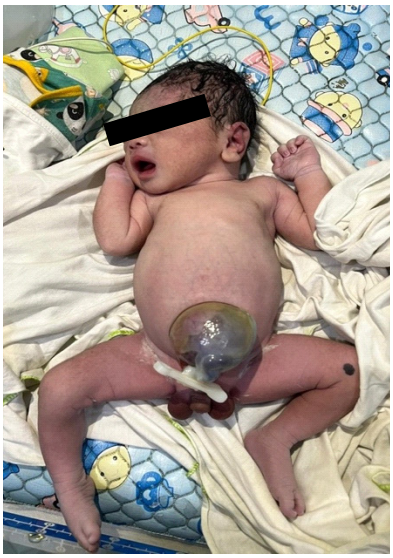

Figure 3. A large omphalocele

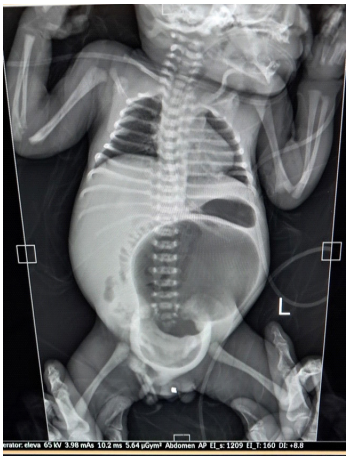

Figure 4. A babygram examination showing distension and dilatation of the rectosigmoid

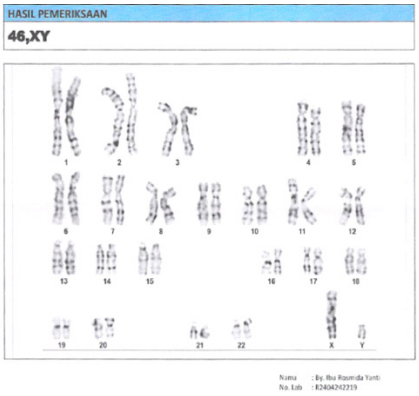

Figure 5. Result of Karyotype analysis

DISCUSSION

The OEIS complex is characterized by a combination of malformations.[1] The initial description of OEIS by Carey et al. was derived from a review of medical records at a large hospital in California, which identified 175 infants presenting with one or more of the aforementioned malformations.[5] In our case, OEI defects were found. The precise cause of the OEIS complex is unknown. However, some cases associated with chromosomal abnormalities, such as deletions in chromosome 1p36 and mutations in homeobox genes. Additionally, maternal factors like diazepam exposure, smoking, obesity, and uterine fibroids have also been linked to certain cases. Some studies states its association with first trimester maternal gonococcal infection and continuous fetal alcohol exposure in utero.[1,6] In our case, however, none of these risk factors were identified.

Its precise etiology remains uncertain, and there is ongoing debate as to whether cloacal exstrophy represents a distinct entity or a point along a developmental spectrum that includes lower urinary tract malformation sequence. One prevailing theory proposes that the anomaly arises from defective development of the lateral mesodermal folds. Insufficient mesodermal reinforcement is thought to cause premature rupture of the cloacal membrane before its fusion with the urorectal septum, ultimately resulting in cloacal exstrophy.[3,4] Alternatively, histopathological findings from some studies suggest that cloacal exstrophy may arise from an early defect in the caudal eminence rather than premature rupture of the cloacal membrane. Despite these hypotheses, ongoing research continues to explore and clarify the embryologic mechanisms underlying cloacal exstrophy.[2]

Prenatal diagnosis is primarily based on ultrasound findings. Early diagnosis is particularly challenging, as the visible findings may be limited.[7] The presence of both defects should raise suspicion for this disorder. Several sonographic features have been identified to help detect this condition. Key diagnostic features include the absence of a bladder, the presence of a persistent cloaca, omphalocele, and myelomeningocele. Additional minor anomalies may involve lower limb deformities, renal abnormalities, intraabdominal free fluid, widened pubic arches, a narrow thoracic cavity, hydrocephalus, and a single umbilical artery. In some cases, a persistent cloaca appears as a cystic mass on the anterior abdominal wall.[8] In our case, only an omphalocele was detected during the prenatal ultrasound examination.

The differential diagnosis should include the exstrophy of cloacal, as one of its hallmark features is the absence of a visible bladder. However, unlike OEIS, bladder exstrophy is generally not associated with the O-I-Ss. Other conditions to consider are the limb-body-wall complex and the pentalogy of Cantrell. Sonographically, limb-body-wall complex may present with thoraco- or abdominoschisis and severe scoliosis. In contrast, pentalogy of Cantrell is characterized by omphalocele accompanied by ectopia cordis, which should be carefully assessed during prenatal ultrasound. Additionally, markedly elevated maternal serum alpha-fetoprotein (MSAFP) levels have been reported in cases of OEIS complex, reflecting the presence of abdominal wall defects and open spinal abnormalities.[9]

Surgical repair of cloacal exstrophy is typically undertaken during the neonatal period, ideally within the first 72 hours after birth. Successful management requires a multidisciplinary team approach, incorporating the expertise of neonatologists, pediatric surgeons, and urologists. Additionally, consultation with neurosurgeons is advised when spinal dysraphism is present to ensure optimal neurological assessment and intervention.[10]

The prognosis can be life-threatening, partly contributes to the high risk of infections and bowel issues. Survival rates for these patients have improved significantly with surgical intervention. Surgical

procedures include repairing the omphalocele, separating the ileocecal connection from the exstrophied bladder plate, and reconnecting an end colostomy while preserving all bowel functions. However, surgical intervention does not always ensure an optimal quality of life. Bowel and urinary incontinence frequently persist following primary repair, primarily due to the underlying neurological defects associated with OEIS. Male patients often face infertility or subfertility later in life, resulting from cryptorchidism and erectile dysfunction.

Although infertility is less commonly reported in females, the presence of a bicornuate uterus has been documented. Advances in reconstructive techniques indicate that undergoing multiple corrective surgeries after the initial repair can improve long-term survival and quality of life.[9]

Identifying OEIS complex prenatally poses a significant diagnostic difficulty, as its clinical features can be subtle and require a highly refined level of vigilance from obstetricians and gynecologists during routine or targeted examinations. A thorough and systematic evaluation must be performed to detect potential malformations in regions such as the lower abdominal wall, urogenital system, lumbosacral spine, and cloacal structures, ensuring that no abnormality is overlooked. It is critical to discern whether each anomaly occurs independently or as part of a cluster of congenital defects involving multiple systems, which may significantly influence both prognosis and planning. The management of OEIS complex demands a well-coordinated multidisciplinary team—including but not limited to fetal medicine experts, pediatric urologists, orthopedic surgeons, and neonatal intensive care professionals—to deliver the best possible outcomes. Given the wide variability in clinical expression and the possibility of unexpected complications, clinicians must prioritize early diagnosis, anticipatory guidance, and flexible care strategies. Integral to this process is maintaining open, continuous communication with the expectant mother and her family, allowing for informed decision-making and emotional preparedness. Thus, the success of care relies on a continuous chain of interventions—starting from prenatal suspicion, through intrapartum management, and into postnatal surgical and supportive therapies—all of which must be executed with unity and precision.

CONCLUSION

This report presents a rare manifestation of the OEIS complex, an uncommon congenital disorder involving multiple systemic malformations of unknown etiology and generally poor prognosis. Prenatal diagnosis relies on sonographic detection of the associated defects; however, confirming the diagnosis remains difficult. Management demands a multidisciplinary approach encompassing both therapeutic and preventive strategies. Considering the significant social impact associated with congenital anomalies, the promotion of genetic counseling, comprehensive prenatal care, early antenatal diagnosis, and timely surgical intervention has become increasingly essential.

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