SEVERE OMPHALOCELE, EXSTROPHY OF THE CLOACA, IMPERFORATE ANUS, AND SPINAL DEFECTS (OEIS): A CASE REPORT

Fahreza L Nasution[1*], Noviardi[2], Sofyan Andri[3], Dhini Ayulie Novri[4], Akbar Husaini Angkat[5]

[1-5]Faculty of Medicine Universitas Riau –Arifin Achmad General Hospital

Email Korespondensi: Fahrezaln@gmail.com

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ABSTRACT

The OEIS complex—comprising Omphalocele, Exstrophy of the cloaca, Imperforate anus, and Spinal defects—is one of the rarest and most severe congenital malformation syndromes, with an estimated incidence of 1 in 200,000–400,000 live births. Early prenatal detection plays a critical role in facilitating appropriate counseling and multidisciplinary perinatal planning. We report a prenatally diagnosed case of severe OEIS complex in a 21-year-old primiparous woman referred for suspected abdominal wall defect and fetal tachycardia. Ultrasound revealed a single live fetus at 32 weeks' gestation with a large omphalocele, protruding bowel loops, and features suggestive of cloacal exstrophy. Postnatal examination confirmed omphalocele, cloacal exstrophy, imperforate anus, ambiguous genitalia, and right-sided congenital talipes equinovarus. These findings were consistent with the classical presentation of severe OEIS. This case underscores the importance of early prenatal diagnosis and coordinated multidisciplinary management in severe OEIS complex. Timely recognition through imaging and delivery planning at a tertiary care center are crucial for optimizing immediate neonatal outcomes. Long-term prognosis remains dependent on the extent of anomalies and access to comprehensive surgical and supportive care.

Keywords: OEIS, Omphalocele, Exstrophy of the Cloaca, Imperforate Anus, and Spinal Defects, Ccongenital Anomaly.

INTRODUCTION

The OEIS complex—an acronym for Omphalocele, Exstrophy of the cloaca, Imperforate anus, and Spinal defects—represents one of the rarest and most severe congenital malformation clusters encountered in fetal and neonatal medicine. This anomaly arises from early embryologic disruption and leads to profound structural abnormalities involving the gastrointestinal, genitourinary, and musculoskeletal systems. The estimated incidence is approximately 1 in 200,000–400,000 live births, making it an exceptionally uncommon condition worldwide, with limited epidemiological data available from Indonesia (Novri et al., 2025; Torres-Cepeda et al., 2021). Due to its complexity and high morbidity, OEIS poses significant diagnostic, perinatal, and long-term management challenges.

Advances in prenatal imaging have allowed earlier recognition of

OEIS. Routine obstetric ultrasonography between 17 and 30 weeks of gestation often reveals hallmark features such as omphalocele, bladder exstrophy, and absence of anal structures, frequently accompanied by additional musculoskeletal or spinal abnormalities. Early diagnosis is crucial as it enables appropriate counseling, referral to tertiary centers, and multidisciplinary planning for delivery and postnatal surgical intervention. However, delayed or missed diagnosis can still occur, especially in low-resource settings where imaging access or expertise may be limited (Dorjey, 2023).

Despite substantial improvements in neonatal resuscitation and reconstructive surgeries, outcomes for infants with OEIS remain highly variable. Prognosis depends on the severity of organ involvement, associated anomalies, and the timing as well as success of staged surgical repair. With coordinated multidisciplinary care, affected individuals may achieve meaningful long-term survival and functional outcomes, though the condition remains life-threatening in its most severe forms. This report presents a rare case of severe OEIS complex diagnosed prenatally, highlighting the diagnostic challenges and perinatal management considerations that accompany this anomaly.

LITERATURE REVIEW

Surgical strategies and outcomes — Contemporary surgical management of cloacal exstrophy and related OEIS defects has evolved toward staged or, in selected centers, single-stage reconstructions aimed at abdominal wall closure, bowel and urinary tract reconstruction, and eventual continence procedures. Recent series report improved survival and functional outcomes with modern perioperative care and specialized reconstructive techniques; nonetheless, long-term morbidity (urologic, gastrointestinal, genital, orthopedic, and psychosocial) remains considerable and requires lifelong multidisciplinary follow-up (Esmaeilizadeh et al., 2023).

RESEARCH METHODOLOGY

CASE REPORT

A 21-year-old primiparous woman referred from other Hospital with ultrasound scanning revealed anomaly of the GI tract positioned outside the abdominal cavity and increased fetal heart rate. She had adhered to a regimen of prenatal vitamins, with no reported history of alcohol consumption or smoking. There was no reported history of congenital anomalies in previous pregnancies, and the current pregnancy had been uneventful until the time of evaluation. Vital signs within normal limits, fetal heart rate examination found 182 beats per minute. There were no sign of labor, leopold examination impression head presentation.

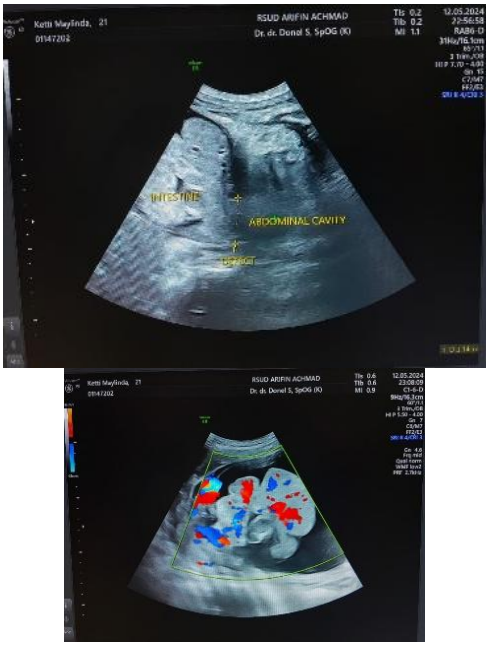

Figure. 1. Ultrasound image showing the defect of the abdominal cavity.

Ultrasound examination revealed a single live intrauterine fetus in cephalic presentation with active fetal movements and a fetal heart rate of 184 bpm. Biometric measurements were consistent with a 32-week gestational age. Further evaluation demonstrated a large omphalocele with bowel loops protruding into the amniotic cavity, while the amniotic fluid volume remained within normal limits. Additional imaging confirmed defects in the lower abdominal wall and suspicion of cloacal exstrophy. Resuscitative measures were attempted intrauterinely due to persistent fetal tachycardia (>185 bpm). Given the severity of anomalies and fetal condition, the team proceeded with emergency cesarean section, following administration of prophylactic antibiotics and consultations with pediatric and anesthesia teams

A live newborn was delivered, weighing 1,500 grams and measuring 33 cm in length. The infant had an APGAR score of 2 at 1 minute and 1 at 5 minutes, indicating severe neonatal distress. The external examination revealed ambiguous genitalia, absence of the anus, and a right congenital talipes equinovarus (CTEV) deformity. Examination of the lower abdominal region demonstrated features consistent with cloacal exstrophy, confirming the diagnosis of OEIS complex.

Following delivery, the newborn was found to have multiple congenital abnormalities consistent with a severe presentation of the OEIS complex. A large omphalocele was evident at the base of the umbilicus, with exposed abdominal contents. The lower abdominal wall defect was accompanied by cloacal exstrophy, in which the bladder and intestinal structures were externally exposed. Perineal examination revealed a complete absence of the anal opening, indicating imperforate anus. Additionally, the infant exhibited a right-sided congenital talipes equinovarus (CTEV), reflecting associated musculoskeletal involvement. Collectively, these anomalies aligned with the classical features

described in severe OEIS cases, supporting the final diagnosis and corresponding with established literature.

Figure. 2. Newborn With Severe OEIS Complex Showing Omphalocele, Cloacal Exstrophy, and Limb Abnormality During Immediate Postnatal Resuscitation.

DISCUSSION

This case presents a severe form of the OEIS complex diagnosed prenatally and confirmed at birth, with classical lesions including omphalocele, cloacal/bladder exstrophy, imperforate anus, and an associated right-sided CTEV. These clinical features from the present case are concordant with the typical OEIS phenotype described in the literature. Epidemiology and embryology — OEIS is an exceptionally rare malformative cluster with reported incidence estimates of roughly 1 in 200,000 to 1 in 400,000 live births, emphasizing its low frequency but high clinical impact. Population-based analyses have established OEIS as a distinct entity arising from early embryologic disruption of infra-umbilical mesodermal structures (Dorjey, 2023).

Prenatal detection and imaging — Advances in prenatal imaging have substantially improved antenatal recognition of exstrophy and OEIS-spectrum anomalies. Over the past two decades, combined use of high-resolution ultrasound and fetal MRI raised prenatal detection rates for OEIS and bladder exstrophy in many series; published analyses report markedly higher recognition of OEIS prenatally compared with earlier eras, with identification commonly between the second and early third trimester. Early antenatal diagnosis permits multidisciplinary planning, parental counseling, and delivery at tertiary centers equipped for immediate neonatal resuscitation and surgical care. he findings in our case (ultrasound showing omphalocele and extruded bowel and planned perinatal consultation) illustrate these practical advantages (Lee et al., 2023; Weiss et al., 2020).

Perinatal management and immediate neonatal care — Newborns with cloacal exstrophy/OEIS frequently require urgent stabilization at birth (airway, thermoregulation, fluid balance, protection of exposed viscera, and antibiotic prophylaxis) and early involvement of neonatology,

pediatric surgery, urology, and orthopedics. The decision-making pathway often balances immediate life-saving goals and timing of definitive reconstruction with the infant's cardiorespiratory stability and associated anomalies. Our management (emergency cesarean delivery, neonatal resuscitation, multidisciplinary consults) corresponds to this recommended perinatal approach (Lee et al., 2023; Weiss et al., 2020).

Prognosis, counseling, and long-term care — Prognosis in OEIS is heterogeneous and depends on the severity of associated defects, timing and success of surgical reconstruction, and access to specialized multidisciplinary care. Recent reviews indicate survival has improved significantly with modern management, shifting the clinical emphasis toward optimizing urinary and fecal continence, sexual and reproductive function, and overall quality of life through lifelong follow-up programs. Families benefit from early, frank counseling about the spectrum of immediate risks, likely staged interventions, expected complications, and requirements for long-term rehabilitative and psychosocial support (Ostertag-hill et al., 2024).

CONCLUSION

Severe OEIS complex is an exceptionally rare congenital anomaly that requires early recognition, coordinated perinatal management, and immediate multidisciplinary intervention to optimize neonatal outcomes. This case highlights the critical role of prenatal imaging and specialized delivery planning in identifying life-threatening malformations and preparing appropriate surgical and supportive strategies. Despite advances in neonatal and reconstructive care, long-term prognosis remains highly dependent on the severity of associated defects and access to comprehensive, lifelong multidisciplinary follow-up.

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