CASE REPORT

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OEIS COMPLEX – A RARE CASE REPORT

ANATOMY

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ABSTRACT

OEIS complex is a spectrum of malformations that result from improper closure of ventral abdominal wall due to failure of convergence of cephalo-caudal and lateral folding's of embryo during 4th week of development with associated defects in development of cloaca and urorectal septum. A third gravida with previous normal obstetric history spontaneously delivered a dead female fetus with multiple congenital anomalies at 32 weeks gestation. The anomalies presented can be grouped under OEIS complex (Omphalocele, Exstrophy of cloaca, Imperforate anus and Spinal anomalies). Diagnosis at birth is made based on the presenting features, but it can be diagnosed prenatally based on the repeated non-visualization of filled bladder, omphalocele, meningocele, club foot etc. it requires long-term multi-disciplinary surgical reconstruction and management to facilitate quality of life.

Key Words: ***Club-foot, Epispadias, Exstrophy of cloaca, Meningocele, Omphalocele***

Introduction

OEIS (Omphalocele, Exstrophy of cloaca, Imperforate anus, Spinal abnormalities) complex represents most severe form of Exstrophy-Epispadias complex (EEC) which includes a spectrum of genitourinary malformations ranging in severity from epispadias (E) to Classical Bladder Exstrophy (CEB) and to Exstrophy of the Cloaca (EC).[1] Exstrophy of Cloaca (EC) is a rare congenital anomaly and is the most severe form of EEC. The first description of a case of OEIS was published by Littre in 1709.[2] The term OEIS complex was first used by Carey et al.,.[3] Subsequently in addition to the four classical manifestations its association with genital, renal and skeletal malformations like spina bifida, symphysis pubis diastasis and limb abnormalities were statistically demonstrated.[4]

Recent studies have suggested that exstrophy of cloaca (EC) and OEIS complex as synonymous entities.[5,6] Reported incidence of OEIS by different authors varies from 1:200 to 4,00,000 live births.[3,4,7] The patient described in the present report had classical malformations (omphalocele, extrophy of cloaca, imperforate anus, scoliosis, and club feet) in addition to absence of gonads and internal genital organs, renal anomalies and gastrointestinal malformations. We hereby present a rare case of OEIS complex – anatomical and radiological findings with embryological explanation.

Case Report

A third gravida of 30 years age with previous normal obstetric history spontaneously delivered a dead female fetus with multiple congenital anomalies at 32 weeks gestation. The weight of the fetus was 2.6 kgs and the crown rump length observed was 20 cms. The fetus was delivered per vagina with a hand prolapse.

Anatomical Observations:

- A defect in the lower anterior abdominal wall (Omphalocele) of 15 x 10 cms size containing liver, spleen and small intestine. The omphalocele presented with ruptured sac (Fig: 1).
- A short umbilical cord of 12 cms attached to apex of Omphalocele (Fig: 1).

- The contents of the Omphalocele were liver with coils of small intestine to its right with esophagus, stomach, spleen, caecum and colon to its left. Duodenum showed features suggestive of atresia and the colon presented with foci of atresia, agenesis and stenosis (Fig: 2).
- The external genitalia presenting as two pairs of folds suggestive of female was observed below the omphalocele (Fig: 3).
- Imperforate anus - In perineal region, there was no anal opening (Fig: 3).
- Lumbosacral meningocele of 8 cms diameter (Fig: 3,4).
- Bilateral club foot (Fig: 1,3)
- Fetal spine showed scoliosis (Fig: 1,4)
- Exstrophy of bladder/cloaca - single ureter and colon opening into the same (Fig: 5)
- On opening the anterior abdominal wall and thoracic cage single kidney, ureter and supra renal were present. Ovaries, uterus and uterine tubes were absent. A foreshortened hindgut was seen opening in to the cloaca. A narrow passage extending from cloaca to urethra was observed (Fig: 5).

Radiological findings: (Fig: 6)

- Spina bifida
- Lumbosacral meningocele
- Scoliosis
- Absence of Iliac bones (bilaterally)
- Invisible Sacro-iliac joints

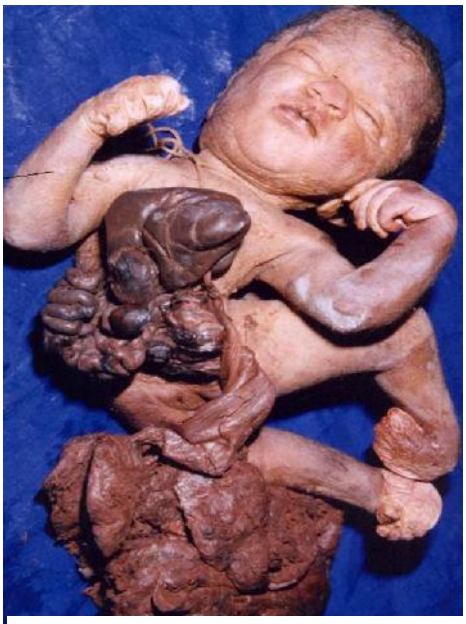


Fig: 1. Omphalocele; A short umbilical cord; Bilateralclub feet and Scoliosis

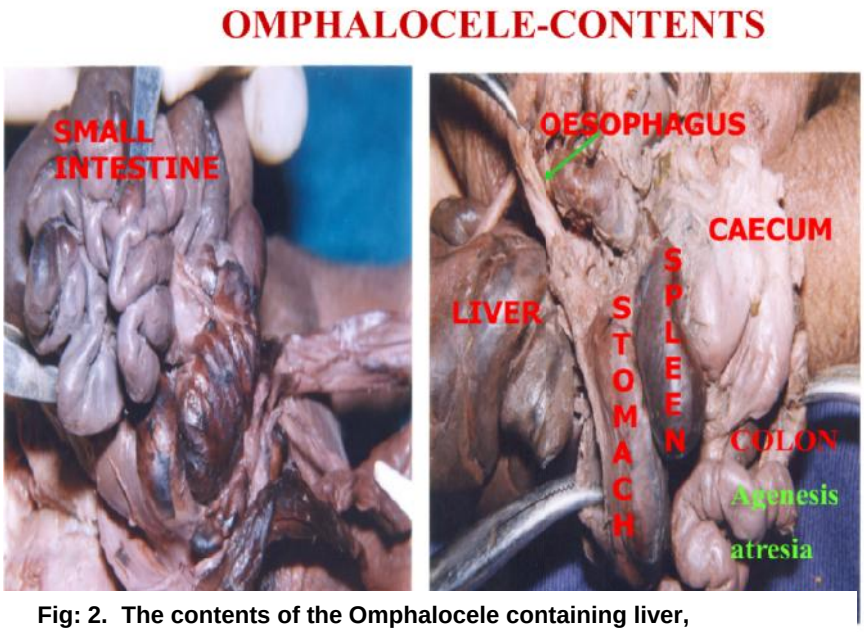
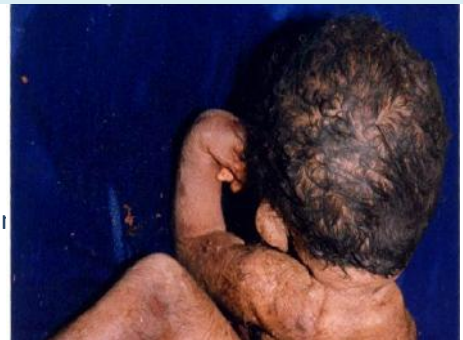


Fig: 2. The contents of the Omphalocele containing liver, spleen, coils of small intestine, oesophagus, stomach, caecum and colon.



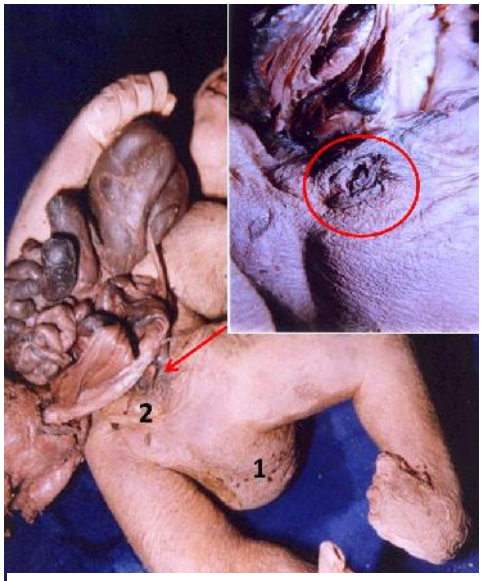


Fig: 3. External genitalia, Imperforate anus, Bilateral club feet. Lumbosacral meningocele.

Fig: 4. Scoliosis. Meningocele

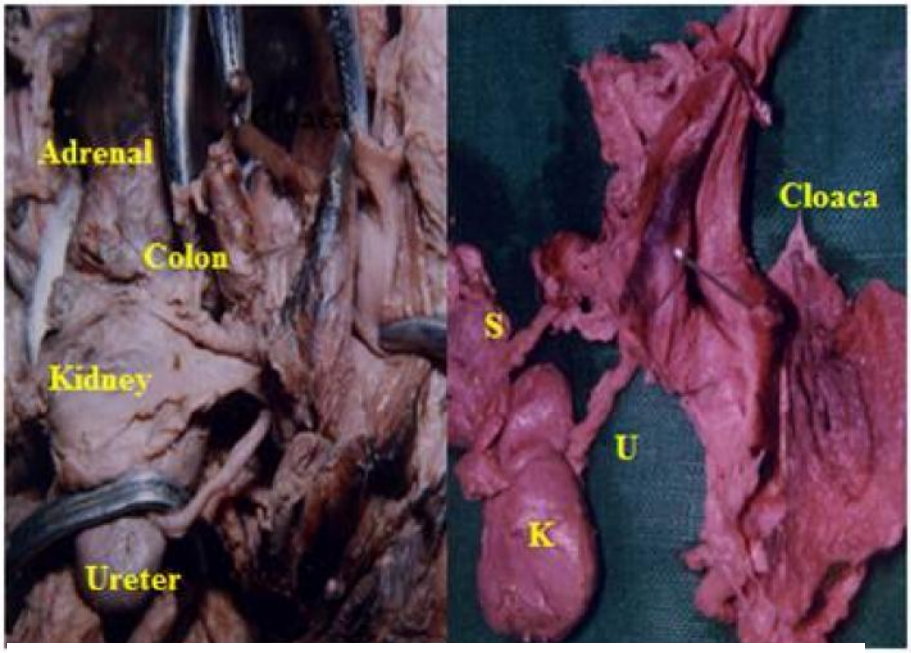


Fig: 5.Exstrophy of bladder/cloaca - single ureter andcolon opening into it. Urethral passage extending from cloaca.

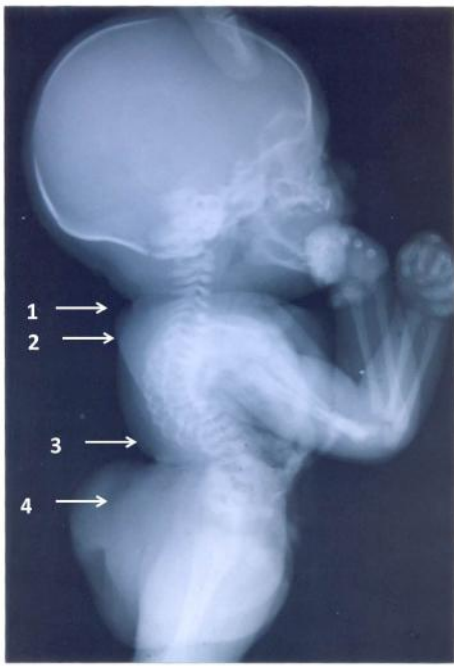


Fig: 6. Spina bifida, Scoliosis, Bilateral absence of iliac bones, Invisible Sacro-iliac joints

Discussion

The classical presentation of cloacal exstrophy consists of omphalocele superiorly with an open plate mucosa inferiorly consisting of two posterior walls of hemibladder on either side with a central strip of intestinal mucosa.[8] These features were observed in the present case but the central intestinal mucosa presented with the opening of extending into the urethral orifice in the external genitalia.

In humans cloaca is a phylogenetic embryonic structure where caudal parts of digestive, genital and urinary systems join. Normal development gives rise to lower abdominal wall with bladder, intestine and anus, genital organs,

pelvic bones, lumbo-sacral spine. OEIS complex results from a single defect of early blastogenesis or a defect of caudal mesodermal migration during primitive streak period that later contributes to the formation of infra umbilical mesenchyme, cloacal septum and vertebrae. These defects lead to improper closure of ventral abdominal wall due to failure of convergence of four ecto-mesodermal folds (a cranial, a caudal and two lateral) of embryo with associated defects in development of cloaca and urorectal septum during 4^{th} week and non-reduction of physiological hernia and non-fixation of gut between 8^{th} -12^{th} weeks of development. Lack of mesoderm in the infra umbilical abdominal wall result in omphalocele. Cloacal exstrophy prevents the development of proctodeum resulting in imperforate anus. Caudal dysgenesis interferes with somite formation resulting in defective vertebrae. [9,10]

The aetiology of OEIS complex was thought to be multifactorial. Higher incidence of OEIS in monozygotic twins than in dizygotic twins suggesting a possible genetic contribution to the occurrence of these defects was reported.[11]

As already described, OEIS complex is difficult to diagnose prenatally and all the abnormalities in the fetus may not be clear until the postnatal examination is completed. [12,13] The major criteria described by the previous investigators for the prenatal diagnosis are Non visualization of fetal bladder, infraumbilical anterior abdominal wall defect, omphalocele and myelomeningocele (MMC). [14,15] The minor criteria are – lower extremity abnormalities (club foot), Renal anomalies, ascites, widened pubic arches, narrow thorax, kyphoscoliosis, hydrocephalus and single umbilical artery. [15,16] In present case all the above criteria major and minor has been observed.

In a study performed by KepplerNoreuil et al.,[17] it has been suggested that the prevalence of omphalocele and OEIS complex may be higher in fetuses with increased nuchal translucency. Possible explanation might be – increased intrathoracic compression due to abdominal defects leading to jugular lymphatic obstruction; Vascular or hemodynamic abnormalities in the early embryonic development. OEIS complex involves abnormalities of almost every system in the body and its clinical presentation shows variations from case to case. [18]

Conclusion:

The anomalous birth of a child embosses strong social stigma over the society. OEIS complex is a situation where compatibility with life is nil or very less. There are no particular etiological factors described in relation to OEIS complex. Prenatal diagnosis relies on sonographic findings. The major sonographic findings include omphalocele, non-visualization of unfilled bladder, persistent cloaca, myelomeningocele, spine and lower limb abnormalities. Hence, regular sonographic evaluation in the pregnant women may create a place for early diagnosis and necessary intervention.

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