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Case Report

Obstetrics and Gynaecology Section

OEIS Complex: A Rare Case Report

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ABSTRACT

The Omphalocele, Exstrophy, Imperforate anus and Spinal defects (OEIS) complex is a rare midline defect that consists of omphalocele, bladder exstrophy, imperforate anus, spinal and genital abnormalities. The presentation varies from case to case representing the most severe form of Exostrophy Epispadias Sequence (EES). Regular prenatal visits and investigations including ultrasound is needed to diagnose this anomaly, but in this case diagnosis was delayed due to irregular antenatal visits by the patient. The aetiology is thought to be multifactorial as was in the present case which was provisionally diagnosed prenatally at 19 weeks. The foetus had classic ultrasound findings of OEIS complex which were later confirmed post expulsion with gross and infantogram features. Advanced level of radiological expertise is required to prenatally diagnose a case of OEIS complex. Depending on severity, management of such cases ranges from termination of pregnancy in severe cases to survival with surgical corrections postnatally in milder forms.

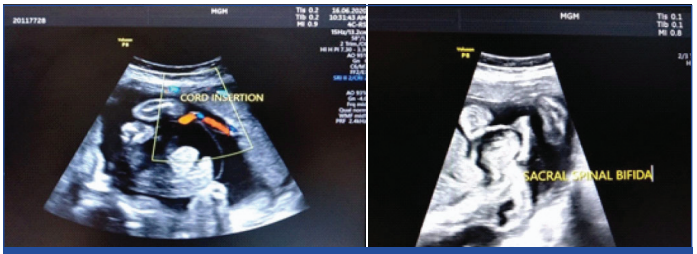
Keywords: Congenital malformation, Exstrophy, Imperforate anus, Omphalocele, Spinal defects

CASE REPORT

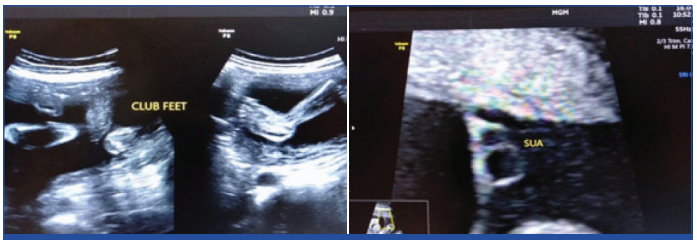
A 23-year-old female, with pregnancy of second gravida with previous missed abortion, married for two years (non consanguinous marriage), conceived spontaneously, came to the Obstetrics Outpatient Department at 19 weeks of gestation to get a second opinion for suspected anomalous foetus where they found presence of a large omphalocoele, kyphoscoliosis and single umbilical artery. Patient had previously only done a viability scan at eight weeks following which she had undergone a 19 week scan directly due to loss of follow-up. There was no history of folic acid intake preconceptionally or in the first trimester. Gestational diabetes mellitus screening was not done. There was a history of missed abortion at 10 weeks gestational age which was managed medically a year ago. Her past and family history was uneventful.

General and systemic examination was unremarkable except for severe pallor. Per abdomen uterus was corresponding to 18 weeks size, relaxed, fetal parts felt. Ultrasound scan findings at our centre revealed large omphalocoele 7.6×6 cm along the midline anterior abdominal wall with herniation of liver, small bowel with overlying membrane [Table/Fig-1], Exstrophy of bladder, Kyphoscoliosis of the lumbosacral region [Table/Fig-2], Bilateral club foot [Table/Fig-3], Single umbilical artery [Table/Fig-4]. The scan findings done at our center were put together and the presence of a rare OEIS complex was diagnosed.

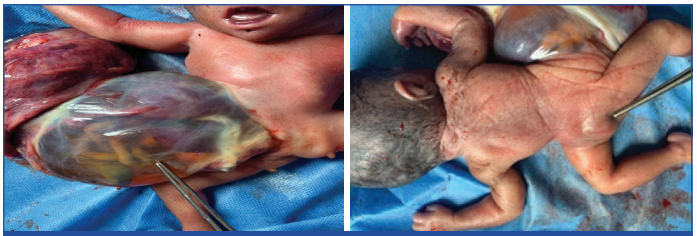
Patient and attenders were counseled regarding poor prognosis of the foetus and they opted for termination of pregnancy. They were also advised further genetic evaluation. Patient was admitted, anaemia was corrected with blood transfusion and with medical induction she expelled vaginally an indeterminate foetus weighing 450 gm. Gross anterior abdominal wall defect was seen extending 5 cm below the xiphisternum upto perineum and abdominal organs protruding as a mass covered by a membrane along with fluid collection omphalocoele to which the placenta was found to be adherent [Table/Fig-5]. Fetal external genitalia was not visualised and cloacal orifice with absence of urorectal septum was noted [Table/Fig-6]. Exstrophy of bladder was seen [Table/Fig-7]. There was kyphoscolisis, closed neural tube defect at sacral region. Imperforate anus and bilateral club foot were among the other congenital defects seen. Rudimentary perineal tags were also seen. Infantogram showed dextroscoliosis with widely separated pubic bones [Table/Fig-8]. Thus, postnatal examination and autopsy



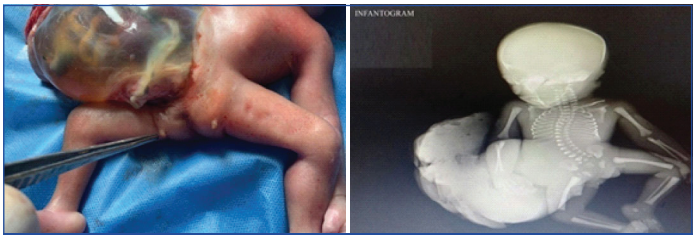
[Table/Fig-1]: Omphalocoele 7.6×6 cm along the midline anterior abdominal wall with herniation of liver, small bowel with overlying membrane.
[Table/Fig-2]: Kyphoscoliosis of the lumbosacral region. (Images from left to right)



[Table/Fig-3]: Bilateral club foot. **[Table/Fig-4]:** Single umbilical artery. (Images from left to right)



[Table/Fig-5]: Gross omphalocoele. **[Table/Fig-6]:** Absence of cloacal orifice. (Images from left to right)



[Table/Fig-7]: Exstrophy of bladder. **[Table/Fig-8]:** Infantogram showing dextroscoliosis with widely separated pubic bones. (Images from left to right)

confirmed the final diagnosis as OEIS Complex (Omphalocoele, Exstrophy of bladder, Imperforate Anus, Spinal Abnormalities).

DISCUSSION

Congenital malformations present with either a major congenital anomaly or a combination of multiple anomalies with similar aetiologies. One such EEC presents a variety of genitourinary abnormalities extending in severity from epispadias (E) to Classical Bladder Exstrophy (CEB) and Exstrophy of Cloaca (EC) often mentioned as OEIS complex has been studied [1]. It was coined by Carey et al., in 1978 and is among the most severe congenital anomaly of the anterior abdominal wall with an incidence of one in every two to four lakh live births making it extremely rare [2,3]. The complex consists of Omphalocele, Bladder exostrophy, Imperforate anus, Spinal and genital abnormalities and is occasionally associated with limb defects also. Various cytogenetics has been suggested in the aetiology of OEIS complex. A possible genetic contribution can be attributed to the increased incidence of OEIS in monozygotic twins than in dizygotic [4]. El-Hattab AW et al., suggested that mutation of a gene located in the 1p36 region could be a cause for OEIS complex [5]. It is a rare anomaly with variability in prevalence by geographic location. There have also been cases of OEIS associated with exposure to diphenylhydantoin and diazepam [6]. Thus, the aetiology of OEIS complex is believed to be multifactorial.

Advanced level of radiological expertise is required to prenatally diagnose a case of OEIS complex. Meizner I et al., and Noack F et al., formulated the prenatal diagnostic criteria for OEIS complex [7,8].

- Major criteria includes infra umbilical abdominal wall defect, non visualisation of urinary bladder, omphalocele, myelomeningocoele and prolapsed ileum appearing as elephant trunk deformity.
- Minor criteria are lower extremity defects, pelvic diastasis and genitourinary anomalies.

Maternal serum alpha-fetoprotein cannot be used to detect all cases of OEIS (as neural tube defects are usually closed type and omphaloceles are covered by thick membrane). Detailed fetal ultrasound examination remains the best tool to detect this condition prenatally. Aneja K studied two cases of OEIS at 12 weeks and 17 weeks of gestation both of which opted for termination of pregnancy emphasising the use of ultrasonography to diagnose and differentiate ventral masses at an earlier gestation showing similar findings and outcome as seen in the present study [6]. Neel N and Tarabay MS reported a case of 10-year-old child with OEIS complex which was detected only after birth and was managed by a multidisciplinary team approach. The child underwent multiple complicated surgical procedures for each defect and the prognosis improved subsequently though, there was high postoperative morbidity [9]. Differential diagnosis includes limb body wall complex, cloacal exstrophy sequence and schisis association although there is a clinical overlap [10]. Management requires initial resuscitation and stabilisation and then based on severity a single staged closure or multi-stage procedure can be done by a multidisciplinary surgical team requiring follow-up throughout life [9].

CONCLUSION(S)

The present case fulfils both major and minor clinical criteria for OEIS complex and in the absence of a typical family history, we presumed that this is an isolated case of probable multifactorial inheritance. Surgical advances and improvement in neonatal care have led to an improved quality of life and a dramatic increase in survival rates. Although the risk of recurrence is very low, it is not impossible. Since foetal ultrasound is the only accurate tool to diagnose the condition, it should be offered in subsequent pregnancies to all parents who had a previously affected pregnancy along with genetic counselling, preconceptional folic acid intake and regular antenatal check-ups to avoid future anomalies.

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