CASE REPORT

Omphalocele, Exstrophy of Bladder, Imperforate Anus and Spinal Defect Complex with Genital Anomalies in a Late Preterm Infant

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A B S T R A C T

Omphalocele, exstrophy of the bladder, imperforate anus and spinal defect (OEIS) complex is a rare congenital multisystemic malformation representing unique anomalies. It was first reported in 1978 through a series of cases with an abnormality of body wall development. We are reporting a case of an infant of 36 weeks gestation, with a family history of consanguinity and oral contraceptive pill intake that was discontinued when the mother was 1-month pregnant. The neonatal examination revealed findings that were consistent with OEIS complex along with the presence of genital anomalies. The infant required multi-staged surgical intervention. We conclude that this case report might illustrate some of the possible risk factors and variability of OEIS complex.

Key words: Cloacal exstrophy, exstrophy of the bladder, omphalocele

ملخص البحث:

تعتبر عقدة الاكشاف المذرقي واحدة من التشوهات الخلقية المتعددة الأجهزة النادرة، وتمثل مجموعة من التشوهات الخلقية المميزة وهي : القيلة السرية، الاكشاف المثاني، والرتق الشرجي وخلل في العمود الفقري. تاريخيا، تم تشخيص أول حالات هذه العقدة في عام 1978 ميلادية من خلال عدد من الحالات التي أظهرت خللا خلقيا في بنية الجدار البطني. يعرض الباحثون حالة خديج في السادس وثلاثين أسبوعا من الحمل ، بتاريخ عائلي من زواج الأقارب، وتناول الأم لحبوب منع الحمل لشهر واحد عند بداية حملها. اثبت فحص الرضيع السريري وجود علامات متماشية مع تشخيص العقدة بالإضافة إلى وجود أعضاء تناسلية ملتبسة . تطلب علاج هذه الحالة إلى تدخل جراحي متعدد المستويات. واستنادا على هذا التقرير، فإننا نخلص إلى أن هذه الحالة النادرة قد توضح بعض عوامل الأخطار المحتملة وتباين عرض عقدة الاكشاف المذرقي.

INTRODUCTION

Omphalocele, exstrophy of the bladder, imperforate anus and spinal defect (OEIS) complex is one of the rarest multisystemic congenital malformations representing unique and characteristic anomalies. It counts for approximately 1:200,000 live births. In 1978, Carey *et al*. reported a series of cases with an abnormality of body wall development known then by many names, most commonly "exstrophy of the cloaca." They proposed the term OEIS complex because the term is simple and recalls all of the defects present.[1] Due to the rarity and lack of comprehensive background of such a diagnosis, our aim is to illustrate some of the possible risk factors and the variable presentations of OEIS complex.

CASE REPORT

A late preterm infant of 36 weeks gestation, product of spontaneous vaginal delivery with Apgar Score of 5 and

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7 at 1 and 5 minutes respectively and admitted initially, was admitted initially as a case of multiple congenital anomalies with genital anomalies.

The infant was born to a 23-year-old Saudi female, G3 P1 + 1, who was not known to have any chronic medical illnesses, but with a history of a previous infant that had died of complicated surgically-corrected diaphragmatic hernia. She was on regular antenatal care follow-up in a private hospital. She had no history of pregnancy-related illnesses or exposure to radiation. There was a history of desogestrel/ethinyl estradiol oral contraceptive pills intake of 3 months duration that was discontinued by the mother who was 1 month pregnant. Regarding the family, there is the first-degree consanguinity between the parents (i.e., cousins), with no similar conditions being reported in the family.

At 28 weeks gestation, an ultrasound study revealed the presence of fetal omphalocele and accordingly she was referred to our hospital for feto-maternal services. The repeated ultrasound scan was of a limited anomaly. Amniocentesis was tried but failed due to advanced gestation.

The delivery went smoothly with the cephalic presentation of the infant and complete placental appearance with three umbilical cord vessels (two arteries/one vein). On examination, the infant looked pale but not cyanosed or jaundiced, with no apparent dysmorphic features. Central nervous system examination revealed an active infant moving upper and lower limbs freely, with myelomeningocele at the lumbosacral area (2 cm × 2 cm) that was covered by intact skin. The cardiovascular and respiratory examination was normal. Gastrointestinal and urinary systems examination revealed omphalocele minor below the umbilicus with an intact sac containing only the bowel. The central area of defect was identified as the caecal plate with a dilated prolapsed terminal ileum with the length of 4–5 cm presenting as characteristic "elephant trunk deformity." There was a small pinhole-sized opening in the proximal ventral aspect of the prolapsed terminal ileum, which noted to be passing the meconium. On lateral aspects, there were exstrophy bladder plates with two hemispheric bladders, each of which contained a visible ureteric orifice. An imperforate anus with an anal dimple was noticed. The genital system examination revealed multiple genital anomalies in the form of asymmetrical widely bifid sacrolabial folds that were hyperpigmented. The gonad was palpable in the left side only with absent phallus and absent urethral opening [Figure 1].

Investigations were performed, including radiographic studies and chromosomal analysis. Echocardiography showed a normal study. The chromosomal study revealed a normal male karyotype of 46, XY with no numerical or structural abnormalities discernible. A computed tomography (CT) scan revealed mildly balanced congenital scoliosis, widening of symphysis pubis indicating an open book pelvic deformity and evidence of congenital bilateral hip dislocation. A multilevel vertebral failure of formation and segmentation with hemivertebra were noted at the mid-thoracic and lower lumbosacral regions. A posterior spinal dysraphism at the lower lumbosacral region was noted that was associated with a low-lying spinal cord that herniates dorsally with skin coverage representing a tethered cord with myelomeningocele [Figure 2].

A multidisciplinary approach had been discussed with a multispecialty surgical team at our center. Due to the absence of a pediatric urology specialty in our hospital, the infant was transferred to a specialized center where the patient underwent a multi-staged surgical intervention to correct the complicated defect.

DISCUSSION

In 1978, Carey *et al*. reported a series of cases with an abnormality of the body wall development. The term OEIS complex was proposed to describe their findings.[1] The description by Carey *et al.* was based on a retrospective search of medical records and identifying 175 infants with one or more of the above-mentioned malformations. Twenty-nine of these infants had two or more of the four cardinal defects and ten infants exhibited the full complex. Among those, genital anomalies were found in six, e.g., absence of external genitalia, ambiguous genitalia, abnormal phallus, epispadias or bifid scrotum. They assumed that these abnormal genital findings were regarded to be secondary features of the complex. The constellation of the complex findings has since then repeatedly been described in the literature and is regarded as an entity.[2] In our case, the cardinal defects of OEIS complex had been identified clinically with the presence of genital anomalies and variable degrees of spinal defects in the form of myelomeningocele, congenital scoliosis and double thoracic hemivertebrae. Based on histopathologic studies in human embryos, OEIS is most likely the result of a very early defect involving the caudal eminence as opposed to an abnormality related to premature rupture of the cloacal membrane. Anatomically, the cardinal findings of cloacal exstrophy include exstrophy of the hemibladders with hindgut extrusion and imperforate

anus. The hemibladders flank the openings of the small intestine, blind-ending large intestine and containing the orifices of ureters and vasa deferentia in males and the uterovaginal canal in females [Figure 3].[3]

Although most cases occur sporadically, there have been several reports of recurrence in siblings suggesting that some cases may have a genetic basis.[4] In our review of the literature, we noted that due to its rarity, most of this complex-related knowledge is still under investigation and reporting, one of the complex's biggest challenges is identifying its related risk factors. We believe that parental consanguinity and use of desogestrel/ethinylestradiol oral contraceptive pills during pregnancy may play a role in the etiology of OEIS complex. In one study, Stoll *et al.* estimated the risk of congenital malformations in consanguineous marriages and found that birth defects in consanguineous couples are 10.3 times more frequent than in offspring of non-consanguineous couples.[5] In another study, Li *et al.* conducted a case–control study of the relationship between oral contraceptive use after conception to the occurrence of congenital urinary tract anomalies (CUTAs) and found that their results were compatible with the hypothesis that oral contraceptive use after conception predisposes the offspring to the development of CUTAs.[6]

The prevalence of OEIS has been estimated at 1:200,000 live births; however, the real incidence is unknown since many cases are incorrectly diagnosed prenatally, or the majority of these pregnancies is terminated or ends with in utero fetal demise.[7]

CONCLUSION

OEIS complex is a congenital multisystemic syndromic entity that should be considered as a medical challenge. Although it was thought to be associated with high mortality risk, this is no longer the case. A crucial modifying step in the management of this complex is its early antenatal diagnosis and detection, where it should be considered as a medical urgency once the diagnosis is reached. A highly specialized multispecialty medical facility is the point of referral where the management must be started initially with stabilizing the patient, then considering either a single-staged or multi-staged surgical intervention. However, given that those patients will have a poor quality of life, such intervention will support the patient's well-being rather than being viewed as active management. As part of our objectives for this case report, identifying unrecognized risk factors will add to the present knowledge of this complex and enhance the treatment

Figure 1: The central area of defect was identified as caecal plate "C" with a dilated prolapsed terminal ileum "I" with the length of 4–5 cm presenting as characteristic "elephant trunk deformity." A small sized opening on the prolapsed terminal ileum which noted to be passing meconium (arrow). On lateral aspects, there were exstrophy bladder plates with two hemispheric bladders "H" each of which contains a visible ureteric orifice "*". Genital anomalies with asymmetrical widely bifid sacro labial folds that were hyperpigmented. The gonad was palpable in the left side with absent phallus.

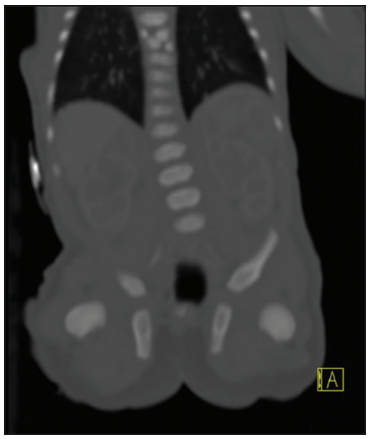

Figure 2: Computed tomography image showing widening of symphysis pubis indicating an open book pelvic deformity with bilateral congenital hip dislocation, mildly balanced congenital scoliosis, multilevel vertebral failure of formation and segmentation with hemivertebra seen at the midthoracic and lumbosacral regions, and posterior spinal dysraphism at the lower lumbosacral region, associated with low-lying spinal cord that herniates dorsally representing myelomeningocele.

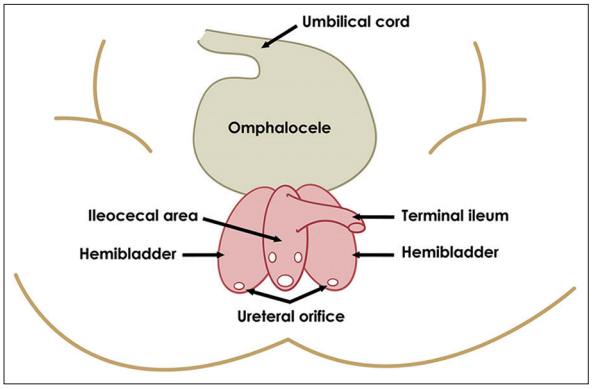

Figure 3: Clinical presentation of cloacal exstrophy in a newborn.

that these patients can be offered. We also conclude that parental consanguinity and use of desogestrel/ethinyl estradiol oral contraceptive pills during pregnancy may play a role in the etiology of OEIS complex.

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Conflicts of interest

There are no conflicts of interest.

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