Exstrophy of cloaca sequence (OEIS complex) with multiple cardiac malformations

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Received - 30 March 2019 Initial Review - 18 April Accepted - 25 June 2019

ABSTRACT

Omphalocele, Exstrophy, Imperforate anus, Spinal defects (OEIS) complex is the most severe birth defect within the exstrophy-epispadias complex. There is exstrophy of the cloaca, failure of fusion of the genital tubercles and pubic rami, omphalocele and incomplete development of the lumbosacral vertebrae with hydromyelia. The diagnosis of OEIS complex mainly relies on sonographic findings. Our case presented with microcephaly, omphalocele, syndactyly, hydromyelia, imperforate anus, single cloacal opening, bifid clitoris, prominent unfused pubic rami and left renal agenesis. In addition, multiple severe cardiac malformations were found on echocardiography. Prognosis is poor when the OEIS complex is compounded by life-threatening malformations. We report a rare case of a preterm neonate presenting with features of OEIS complex with multiple cardiac malformations.

Keywords: *Cardiac malformations, Exstrophy, Imperforate anus, Omphalocele, Spinal defects.*

Omphalocele, Exstrophy, Imperforate anus and Spinal defects (OEIS) complex was first described in 1978 by Carey et al. It is a rare congenital anomaly with multi-system involvement affecting 1 in 400,000 births [1]. The incidence may be underestimated because the condition may not be identified in stillborns. The development of the cloacal membrane occurs between days 5 and 42 of gestation. During the 8 to 16 mm stage, the urorectal septum grows to divide a single cloacal chamber into the anterior urogenital system and the posterior alimentary system [2].

OEIS complex is caused by a single localised defect of the mesoderm during early development (Fig. 1). The consequences are persistence of a common cloaca into which the ureters, ileum and hindgut open with exstrophy of the cloaca, failure of fusion of the genital tubercles and pubic rami, omphalocele and incomplete development of the lumbosacral vertebrae with herniation of grossly dilated central canal of spinal cord-hydromyelia, yielding a soft, cystic, skin covered mass over the sacral area, sometimes asymmetric in its positioning [3].

Surgical intervention has improved the prognosis in OEIS complex. However, the outcome depends on the presence of other life-threatening malformations. The association of multiple cardiac malformations with OEIS complex is rare. We present a case of a preterm neonate with OEIS complex and multiple cardiac malformations.

CASE REPORT

A female neonate was born of non-consanguineous marriage to a 24 years old primigravida at 34-weeks gestation by emergency LSCS. The baby cried immediately after birth with an Apgar score of 8/10 at 1 min and 9/10 at 5 min and was transferred to NICU for prematurity and low birth weight (LBW) with multiple congenital anomalies.

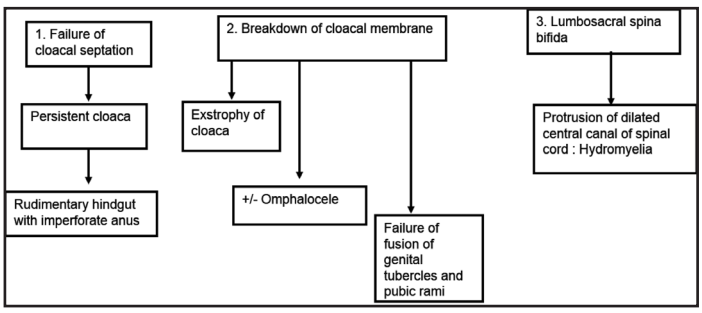

Figure 1: Early defect in mesoderm leading to formation of OEIS complex

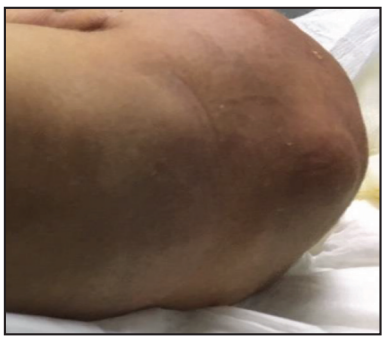

Figure 2: Sacral swelling – Hydromelia

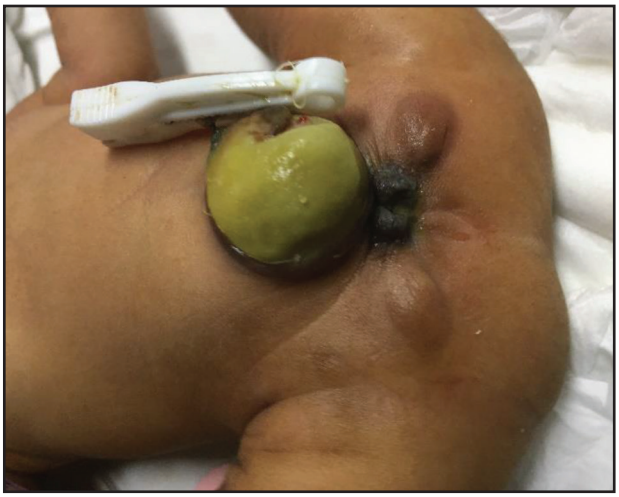

Figure 3: Omphalocele, anterior abdominal wall defect, imperforate anus, single cloacal opening, bifid clitoris and prominent unfused pubic rami.

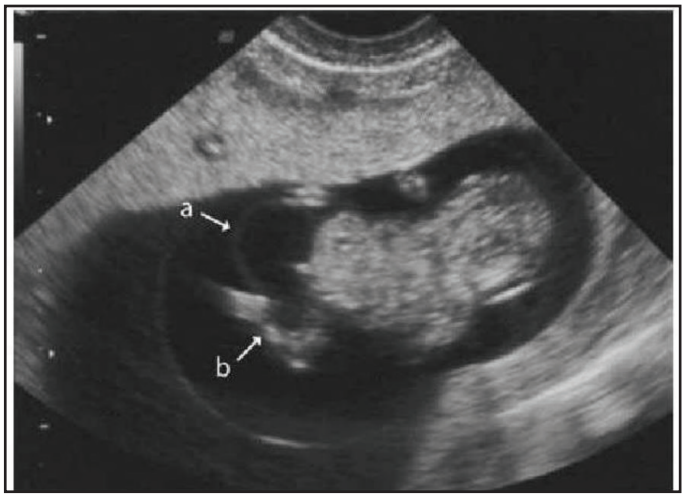

Figure 4: Antenatal USG showing soft well-defined mass in the anterior abdomen suggestive of omphalocele.

Baby's length was 42 cm, weight 2.2 kg and head circumference 29 cm (Z score less than -3SD). Physical examination showed microcephaly, omphalocele, syndactyly, sacral swelling-hydromyelia (Fig. 2), anterior abdominal wall defect, imperforate anus, single cloacal opening, bifid clitoris and prominent unfused pubic rami (Fig. 3). Hypotonia and hyporeflexia were present in the lower limbs.

Antenatal ultrasonography (USG), done at 31 weeks, showed an omphalocele (Fig. 4). The umbilical cord showed a single umbilical artery. The fetal head and rest of the spine were normal. USG abdomen-pelvis and local USG done postnatally showed an absence of the left kidney and gall bladder and the presence of a moderately large omphalocele. Local USG of the sacral region showed a cystic structure (hydromyelia) measuring 1.4cm X 1.8cm X 2.6cm in the presacral region with multiple septa within. The overlying skin was intact. Chest X-ray showed a boot shaped heart (Fig. 5). 2D echocardiography showed Tetralogy of Fallot (TOF), pulmonary atresia, confluent pulmonary artery stenosis and moderate-sized patent ductus arteriosus (PDA) (Fig. 6). Karyotyping was normal.

The baby was admitted in NICU in view of prematurity and LBW with multiple congenital anomalies and was on oxygen by prongs. The baby's condition deteriorated and was intubated after 4 hours and kept on pressure control ventilatory mode and was started on IV antibiotics. The baby died on the second day of life due to multiple congenital anomalies.

DISCUSSION

OEIS complex is a rare genetic disorder characterised by the presence of multiple congenital anomalies including spinal, rectovesical system, limb, anterior abdominal wall, and renal anomalies [1,3]. Major sonographic findings include non-visualisation of the bladder, persistent cloaca, omphalocele, and myelomeningocele [4,5]. Almost all of these features are found in our case.

OEIS complex involves multiple systems and shows variability from case to case [6]. The etiology of OEIS complex is thought to be multifactorial. It may be related to teratogen exposure in the early stage of pregnancy. Although most cases occur sporadically, a case with family history and recurrences in

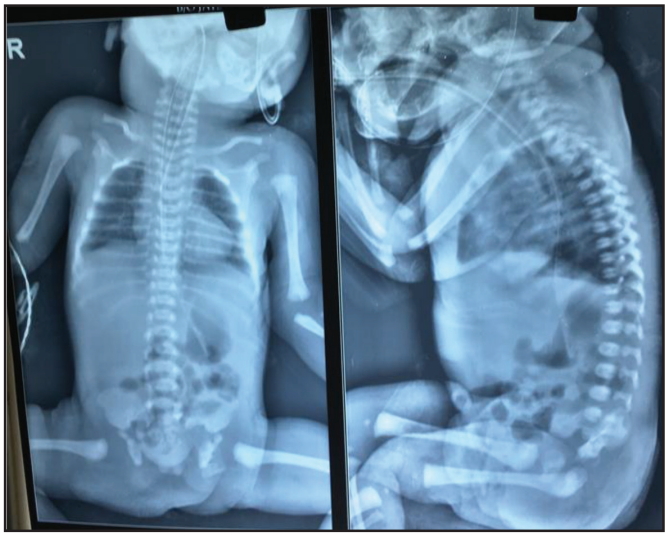

Figure 5: X-ray showing boot shaped heart

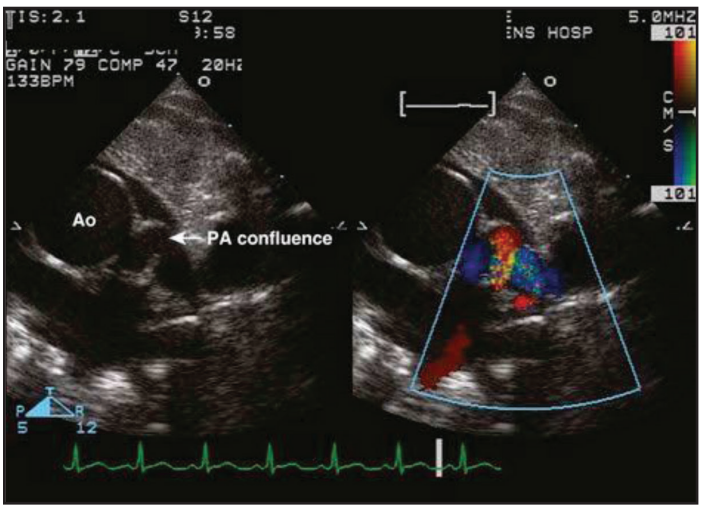

Figure 6: 2D echo showing Tetralogy of Fallot, pulmonary atresia, confluent PAS, moderate sized PDA

siblings has been reported. OEIS has also been identified in one fetus of a twin gestation [7,8]. Gastrointestinal malformations are found in almost all cases. The most prominent is a blind-ending colon with imperforate anus. Other anomalies include duplication of the colon, intestinal malrotation, atresia, short gut, and situs inversus. Imperforate anus was present in our case. Ambiguous genitalia is common in OEIS [9,10,11].

In cloacal exstrophy, the persistent cloacal membrane impedes the migration of the mesoderm streak to form the anterior abdominal wall. The persistent cloacal membrane is unstable and will rupture. If it ruptures before the division of the cloacal chamber by the urogenital septum, cloacal exstrophy occurs. If it ruptures after the formation of a urogenital septum, the result will be exstrophy of the bladder [3,12]. Bladder exstrophy is identified in almost all the patients and was also seen in our case. Renal agenesis has been reported and was present in our case. Other renal anomalies, which include rudimentary kidney, pelvic ectopic kidney, ureteropelvic junction obstruction, malrotation, and crossed renal ectopia, were not seen in our case [11].

A variety of skeletal anomalies have been documented. Majority of patients present with pubis diastasis as seen in our case. The incidence of spinal dysraphism is 100%, as revealed by magnetic resonance imaging. Common spinal abnormalities include hemivertebrae, tethered cord, and lipomyelocystocele, extra vertebrae, and absent vertebrae. Our patient had hydromelia as commonly reported.

Limb anomalies may be secondary to defects of the spinal cord. Arthrogryposis, talipes, syndactyly, and thumb hypoplasia have all been documented in patients with this condition [13]. Although central nervous system anomalies are not very common, Chiari malformation and hydrocephalus may be present in patients with open spinal defects but not seen in our case. Cardiac malformations are usually not associated with OEIS complex. However, our neonate presented with multiple cardiac anomalies such as TOF, pulmonary atresia, confluent PAS and moderate-sized PDA in addition to the OEIS complex.

Batra P et al reported a similar rare case of OEIS complex associated with multiple cardiac malformations [14]. Another case report by Kant SG et al described severe cardiac defects in a patient with the OEIS complex [15]. Källen K et al conducted a population study for the analysis of infants with multiple malformations and found 194 probable OEIS complex cases among 5,260 infants with multiple malformations. However, no specific association with cardiac defects was apparent [12]. Carey JC et al discussed the typical malformations present in babies with OEIS complex [1]. Girz BA et al described the first-trimester sonographic findings associated with OEIS complex and reviewed the literature regarding this rare congenital anomaly [4].

The differential diagnosis includes Pentalogy of Cantrell and Limb Body Wall Complex [3]. Pentalogy of Cantrell is a rare disorder characterised by omphalocele, ectopia cordis, diaphragmatic defect, pericardial defect or sternal cleft, and cardiovascular malformations. It results from abnormal migration of the sternal anlage and myotomes in the early embryonic stage. Limb Body Wall Complex is a rare condition characterised by anterior thoracoabdominal wall defect and involves abdominoschisis usually large and left-sided and almost always present with limb defects, neural tube defects, abnormalities of urogenital organs and is incompatible with life.

CONCLUSION

Isolated OEIS complex has shown excellent survival rates following surgical repair. Additional malformations associated with OEIS complex make this condition life-threatening. Prenatal diagnosis is dependent on the sonographic visualisation of these multiple defects; however, the diagnosis remains challenging. Awareness of the severity of this anomaly and subsequent poor quality of life for individuals born with OEIS complex with other life-threatening malformations may lead patients to elect for medical termination of pregnancy. Morbidity and mortality are dependent on the presence and severity of other malformations.

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Funding: None; Conflict of Interest: None Stated.

How to cite this article: Sadula SR, Kanhere SV, Phadke VD. Exstrophy of Cloaca Sequence (OEIS Complex) with Multiple Cardiac Malformations. Indian J Case Reports. 2019;5(4):317-319.

Doi: 10.32677/IJCR.2019.v05.i04.006