Omphalocele, exstrophy cloacae, imperforate anus, and spinal defects complex with absent foot

Rinku Saini[1], Priti Saini[2], Lovesh Saini[3], R N Sehra[4]

From Departments of [1]Pediatric Medicine, [2]Pathology, [3]Preventive & Social Medicine, and [4]Neonatal Intensive care Unit, Mahila Chikitsalaya, Department of Pediatric Medicine, Sawai Man Singh Medical College, Jaipur, Rajasthan, India

Correspondence to: Dr. Rinku Saini, D-3, Doctors Flats, Gangwal Park, Jaipur – 302 004, Rajasthan, India. Phone: +91-9001986353. E-mail: dr.rinkusaini@gmail.com

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Omphalocele, exstrophy cloacae, imperforate anus, and spinal defects (OEIS) complex is an extremely rare congenital disorder and the most serious form of abdominal midline malformation [1-3]. The characteristic defects of OEIS complex involve the urinary system, musculoskeletal system, the pelvis and pelvic floor, abdominal wall, genitalia, spine, and anus. It covers a spectrum with different severity levels ranging from epispadias representing thess mildest form to the full picture of classical bladder exstrophy and exstrophy of cloacae, also referred to as OEIS complex - the most severe form [1-5]. Incidence varies from 1/200,000 to 400,s00 live births [1]. It results from the premature rupture or abnormal development of the cloacal membrane. The timing of the rupture may determine the severity within the spectrum [3-5]. Over the past 40 years, the focus has shifted from survival to improving patient outcomes and ensuring ths optimum quality of life. The priority being given to urinary, gastrointestinal, and genital reconstruction designed to adapt the patient as a free person and of appropriate gender [1].

A preterm (34–36 weeks) baby with a birth weight of 1.9 kg was born to a third gravida mother aged 25 years by normal vaginal delivery at Mahila Chikitsalaya, SMS Medical College, Jaipur. First was a 6-year-old male, alive and healthy, second was an abortion in the 3rd month of pregnancy, and third was the index case. Antenatal history was uneventful. The antenatal period was improperly supervised with no antenatal ultrasounds, no history of any drug intake or gestational diabetes. The delivery was uneventful with an immediate cry after birth, Apgar score of 7/10 at 1 min after birth. The baby, soon after birth, was admitted in a neonatal intensive care unit, of this hospital. On examination, the vitals were stable, and clinically, the baby had an omphalocele, bladder exstrophy with no discernible external genitalia, anal atresia, and absent right foot (Fig. 1). Sex could not be determined. The infant gram was suggestive of hemivertebrae and pubic diastasis (Fig. 2).

A diagnosis of OEIS complex was made on the basis of these findings and baby was immediately shifted to pediatric surgery department. In the Stage 1 operation, abdominal wall and bladder closure were done and the baby was discharged with a colostomy *in situ*. The genetic analysis of the baby could not be done due to the non-affordability of the parents. The baby gradually became dull at home, stopped accepting feeds, and expired about a week after discharge. The cause of death might be sepsis due to poor care at home. The postmortem of the baby was not carried out.

OEIS complex, also known as cloacal exstrophy, is the most severe birth defect within the exstrophy-epispadias complex. It is characterized by omphalocele, exstrophy of cloacae, imperforate anus, and spinal defects [1-4]. A baby with this condition will have the bladder and a portion of the intestines exposed outside the abdomen with the bony pelvis open like a book [2]. Etiology remains unknown. The reported risk factors include young maternal age, increased parity, and *in vitro* fertilization. The possible role of genetic factors is based on the increased recurrence risk for offsprings of affected individuals [6-8]. Among siblings, the recurrence risk is 1% in non-consanguineous and non-affected parents. As for other birth defects, a small subgroup may follow Mend Elian inheritance, whereas in the majority, it is inherited as a complex trait with multiple genetic factors and complex gene-gene or gene-environment interaction contributing to its formation [6].

Besides the characteristic anomalies, other reported associated anomalies include [9] Meningocele or myelomeningocele, absent or low-lying kidneys, hydronephrosis, and hydroureter, orthopedic anomalies such as clubfoot deformities, absence of feet, tibial or fibular deformities, and hip dislocations and vascular anomalies like reduplication of aorta. The association of absent foot with OEIS complex is frequent and present in 30–79% of the cases [10].

The diagnosis is made by the characteristic clinical picture [1-3]. There are no specific laboratory tests; although it can be diagnosed antenatally by ultrasound. The very typical clinical picture does not implicate any other differential diagnosis. The management is primarily surgical, undertaken as a multi-staged approach with an aim to achieve secure abdominal wall closure, urinary continence with preservation of renal function and adequate cosmetic and functional

Figure 1: Omphalocele with attached umbilical cord, bladder exstrophy, imperforate anus, no discernible, external genitalia, and absent right foot

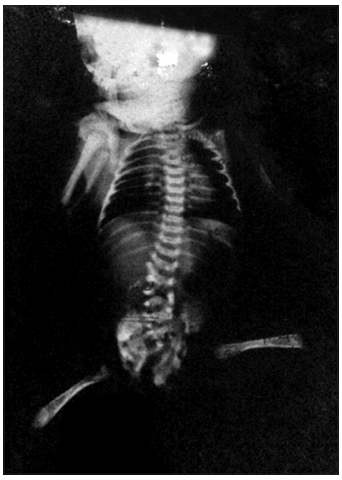

Figure 2: Skiagram showing hemivertebrae with spina bifida and pubic diastasis

genital reconstruction. OEIS complex remains a rare and challenging diagnosis [1,6]. Advances in the medical and surgical management have allowed for improved survival and continence rates, but still, the overall prognosis remains guarded and these children, even with the best care, require lifelong support.

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