Patel V et al. GAIMS J Med Sci 2023;3(1) (Jan-Jun):29-32
Online ISSN:2583-1763

Case Report

Omphalocele, Exstrophy of Cloaca, Imperforate Anus and Spinal Defect – An Autopsy Report

Vipa Patel [1], Kinalee Chothani [1], Jaimin Patel [1], Akansha[2], Dhaneshwar N Lanjewar [1]

1-Department of Pathology, Gujarat Adani Institute of Medical Sciences and GK General Hospital, Bhuj, Kachchh, Gujarat-370001

2-Department of Obstetrics and Gynecology, Hind Medical Institute of Medical Sciences, Barabanki, Lucknow.

* Correspondence: Dr Vipa Patel (vipapatel94@gmail.com)

ABSTRACT

Cloacal exstrophy is a rare condition that occurs during the prenatal development of the lower abdominal wall. Cloacal exstrophy is also known as OEIS Syndrome because of the four features that are typically found together: Omphalocele, Exstrophy of the bladder and rectum, Imperforate anus, Spinal defects.[1] We describe a case of 29-year-old female with 3 months' gestation, her USG showed rudimentary right lower limb and anterior abdominal wall defect for which pregnancy was terminated and fetal autopsy was performed. This is a first case report from our institute in which the fetal autopsy finding showed features of OEIS syndrome.

Keywords: OEIS, Cloacal exstrophy, Omphalocele, Imperforate anus, Spinal defect

INTRODUCTION

Omphalocele, Exstrophy, Imperforate anus and Spinal defects (OEIS) complex was first described in 1978 by Carey et al.[2] It is a rare congenital anomaly with multi-system involvement affecting 1 in 200,000 to 400,000 births. There is no obvious etiology while, sporadic nature being the commonest in most of the cases. Besides the clinically recognized classic malformations, it can be variably associated with spina bifida, genital abnormalities, renal abnormalities, symphysis pubis diastasis, and limb abnormalities.[3] The defect occurs due to embryonic failure of convergence of mesodermal migrations and development of urorectal septum.[2]

CASE REPORT

A 29-year-old female with G3P1A1L1 came to obstetric OPD with 3 months of gestation. She had 8-year-old female child with full term normal vaginal delivery. Her second pregnancy was terminated at 5 months of gestation due to USG reporting of congenital anomaly. In current pregnancy USG finding showed rudimentary right lower limb and anterior abdominal wall defect for which patient and her family were counselled and family opted for termination of pregnancy. The amniotic fluid chromosome analysis showed 46XX karyotype. There was no significant antenatal history of exposure to teratogenic drugs, infection, diabetes, hypertension and exposure to radiation. Fetal autopsy was performed to know congenital anomalies.

Autopsy findings:

The fetus was 15 cm in length it had small head, short neck and showed bilateral proptosis and the ears were normal. The right lower limb was rudimentary and was 1 cm in length and left lower limb was 5 cm in length. Both hands had 5 fingers each and lower extremity had 5 toes each. The abdomen was distended and anterior abdominal wall showed a exophytic cystic structure attached to umbilical cord. Distended abdomen contained transparent sac of 1.5 cm in diameter, it showed fragments of reddish liver tissue. [Figure-1] The clitoris was enlarged, there was no meatal opening and the anus was imperforate. [Figure-2]
The omphalic sac was attached to distended urinary bladder and the distal end of the urinary bladder was attached to dilated colon (vesico-colon fistula) and the terminal end of the colon was blind. [Figure-3]

Radiological study after post-mortem examination of the complete fetus was carried out. The right limb did not show bones and the vertebral column was normal, there was no spina bifida. [Figure-4]

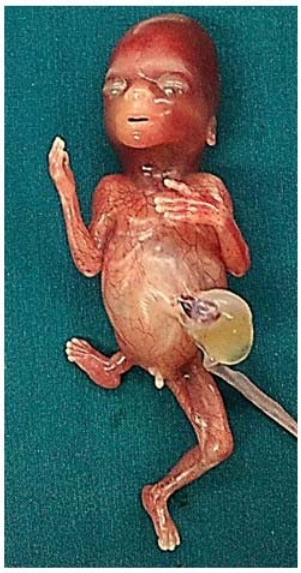

Figure 1: Shows omphalocele containing fragments of liver rudimentary right limb and enlarged clitoris

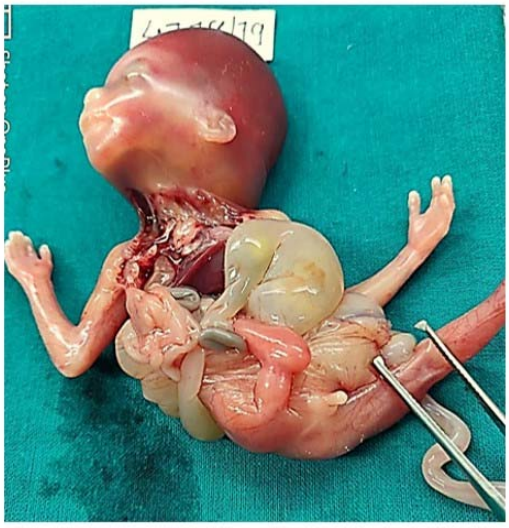

Figure 2: Shows distended urinary bladder and colon and imperforate anus

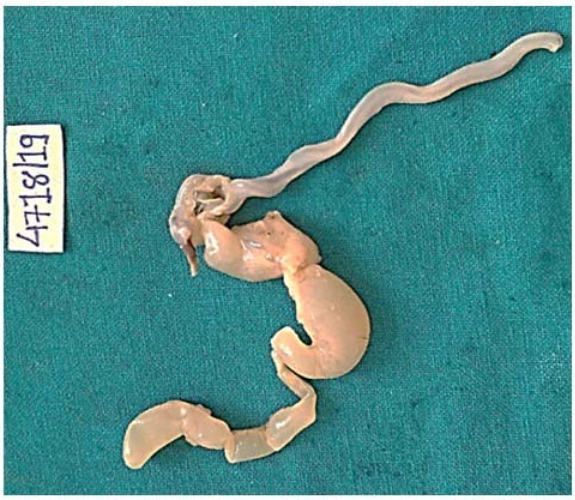

Figure 3: Shows umbilical cord attached to omphalocele attached to urinary bladder, attached to dilated colon and blind end of the rectum

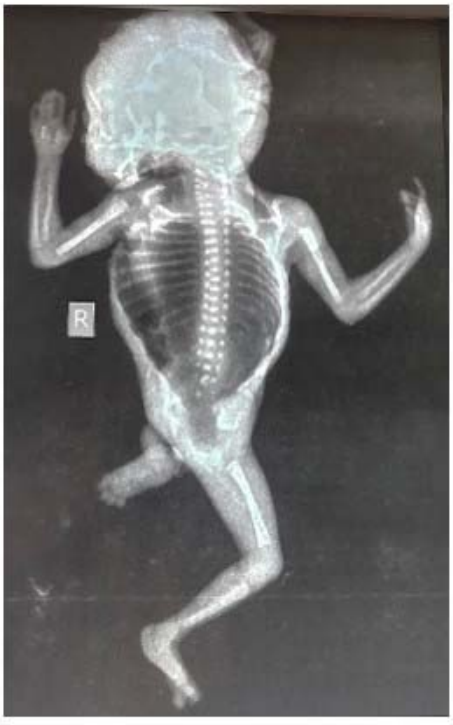

Figure 4: Whole body X-ray showing of fetus with normal spine and absent bones in right lower extremity

The heart was small in size and its anterior surface was not covered with lungs and both lungs were small in size. The thymus was in normal anatomical position and there was no diaphragmatic hernia. Liver was enlarged and was occupying right and left hypochondrium, the spleen was absent. The left kidney was small than right kidney and the ureters were absent. The cut surface of both the kidneys showed numerous cysts. The brain was normal and the placenta did not show any significant finding. The urinary bladder on histology showed transitional lining and colon showed lining of columnar epithelium. The histology of hypoplastic lung showed absent alveolar development. The histology of both kidneys showed large cysts lined by flattened cuboidal epithelium and an intervening disorganized fibrotic parenchyma with islands of cartilage and rare glomeruli and immature tubules (Cystic renal dysplasia).

DISCUSSION

In human 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, anus, genital organs, pelvic bones and lumbo-sacral spine. OEIS complex results from defect in early blastogenesis or a defect of caudal mesodermal migration during primitive streak period that later contributes to the formation of infraumbilical 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 4th week and non-reduction of physiological hernia and non-fixation of gut between 8th -12th 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.[4]

It results from developmental defects affecting the mesenchyme which is required for the development of infraumbilical mesoderm, the urorectal septum and lumbosacral somites.[5]

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 siblings has been reported and OEIS has also been identified in the fetus of a twin gestation.[6]

In some patients with OEIS complex trisomy 21, trisomy 18, triple X syndrome, turner mosaicism and 1p36 deletion is found. however, given the relatively high prevalence of these chromosomal disorders in the general population, it is unlikely that OEIS complex is associated with these chromosomal aberrations.[7]

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. Ambiguous genitalia are common in OEIS.[8]

Prenatal diagnosis of OEIS complex can be done by anomaly scan between 18 to 22 weeks of gestation based on following features, non-visualization of fetal bladder, infra umbilical anterior wall defect, omphalocele, myelomeningocele and, minor criteria like lower extremities anomalies, ascites, widened pubic arches, kyphoscoliosis,hydrocephalus and single umbilical artery. [9]

Compatibility of life with OEIS complex is nil or very less. Even if baby survives it requires multiple surgeries with many potential complications including renal, reproductive, ambulatory impairments and psycho social defects. In absence of open neural tube defects brain is structurally and functionally normal and normal cognitive development can be expected.[10]

A review of the English literature in online archival database up to 2021 was performed. We found 31 Indian case report about OEIS syndrome.

CONCLUSIONS

Cloacal extrophy is a rare condition, the exact etiology is not known, and early amnion rupture is common cause. Chromosomal studies are usually normal and recurrence is generally low. Treatment varies from appropriate perinatal management to termination of pregnancy depending on the severity. 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. Medical and surgical management of these children have improved survival rate.

REFERENCES

1) Rajgopal L, Bhosale Y, Nandanwar YS. Omphalocoele, exstrophy of cloaca, imperforate anus and spinal defects (OEIS complex) – a case report and review of literature. Anat Soc India 2007; 56:41-3.

2) Carey JC, Greenbaum B, Hall BD. The OEIS complex (omphalocele, exstrophy, imperforate anus, spinal defects). Birth Defects Orig Artic Ser. 1978; 14:253-263.

3) Bohring, A. OEIS complex, VATER, and the ongoing difficulties in terminology and delineation. Am. J. Med. Genet., 202;107:72–76.

4) Subhatra Devi V, Md. K. Faheem N, Vidyavathi Ch. Usharani V. OEIS complex: a rare case report. Int J Res Dev Health. 2013; 1:21-5.

5) El-Hattab AW, Skorupski JC, Hsieh MH, Breman AM, Patel A, Cheung SW, et al. OEIS complex associated with chromosome 1p36 deletion: A rare case report and review. Am J Med Genet A. 2010;152A:504-11.

6) Feixue Y, Charlotte H, Armando F. OEIS Complex a Case Report. Journal of Diagnostic Medical Sonography. 2007; 23:13-18.

7) Vlangos CN, Siuniak A, Ackley T, van Bokhoven H, Veltman J, Ram Iyer, Park JM, Keppler-Noreuil K, Keegan CE. Comprehensive genetic analysis of OEIS complex reveals no evidence for a recurrent microdeletion or duplication. Am J Med Genet Part A 2011; 155:38–49.

8) Cloacal exstrophy. Urology Care Foundation. 2016.Available at: http://www. urologyhealth.org/ urologic-conditions/cloacal-exstrophy [Accessed on 19 December 2022]

9) Noack F, Sayk F, Gembruch U. Omphalocele exstrophy imperforate anus defects complex in dizygotic twins. Foetal Diagn Ther. 2005; 20:346-8.

10) Tiblad E, Wilson D, Carr M, Flake A, Hedrick H, Johnson M, et al. OEIS sequence: A rare congenital anomaly with prenatal evaluation and postnatal outcome in six cases. Prenatal Diagn. 2001; 28:141-7.

Source of support: Nil

Conflict of interest: None declared

How to cite: Patel V, Chothani K, Patel J, Akansha, Lanjewar D N. Omphalocele, Exstrophy of Cloaca, Imperforate Anus and Spinal Defect – An Autopsy Report.GAIMS J Med Sci 2023;3(1):29-32.
https://doi.org/10.5281/zenodo.7673714