Perinatal Autopsy Finding in OEIS Complex Associated with Other Congenital Anomalies

Asaranti Kar¹, Tushar Kar², Ipsita Dhal³, Priyadarshini Biswal⁴, Satyajit Jena⁵

SCB Medical College Campus, Cuttack, 753007, India

Abstract: OEIS complex is one of the congenital anomalies not compatible with life. It comprises of omphalocele, exstrophy of the cloaca, imperforate anus and spinal defects with incidence of 1:100,000 live births. It can be associated with other anomalies of urinary tract, genital tract, skeletal defects vari ably. Diagnosis can be done prenatally by ultrasonography. We present a case of OEIS complex, cloaca, imperforate anus and spinal defects with incidence of 1:100,000 live births. It can be associated with other anomalies of urinary tract, genital tract, skeletal defects vari ably. Diagnosis can be done prenatally by ultrasonography. We present a case of OEIS complex with other associated congenital anomalies like imperforate anus, absent external genitalia, gonads, appendix with single kidney, meningocele and limb deformity in the form of talipesequinovalgus in a 29 weeks foetus. It was delivered vaginally by a fifth gravida female without any risk factors and all the four siblings are leading normal post-natal life.

Keywords: Imperforate anus, OEIS complex, talipesequinovalgus, omphalocele. cloacalexstrophy

1. Introduction

The OEIS complex comprises a combination of defects including omphalocele, exstrophy of the cloaca, imperforate anus, and spinal defects. It is exceedingly rare. Recently a prevalence rate of 1:100,000 live births have been reported by Keppler-Noreuil in 2007. [1] Most recent reports indicate a male-female sex ratio of 2:1. It represents most severe form of Exstrophy-Epispadias complex (EEC) which ranges in severity from epispadias to exstrophy of cloaca. It has been suggested by some authors that OEIS and exstrophy of cloaca are synonymous entities. [2,3]There is no obvious etiology in most of the cases. The few reported patients with family members having similar malformations or with chromosomal anomalies suggest a genetic contribution in few non sporadic cases. Other reports have linked cloacalexstrophy to certain exposures, twinning and in vitro fertilization. Besides the clinically recognised classic malformations, it can be variably associated with spina bifida, genital abnormalities, renal abnormalities, symphysis pubis diastasis, and limb abnormalities.[4] The patient described in the present report had classical malformations (omphalocele, exstrophy of cloaca, imperforate anus and scoliosis) in addition to absence of gonads and internal genital organs, single kidney, absent appendix and club foot. We hereby present a rare case of OEIS complex with detail autopsy findings and associated with other congenital abnormalities.

2. Case Report

A 42 year old fifth gravida with previous normal obstetric history and last child birth 22 years back came with complaints of amenorrhoea and abdominal distension since 5 months. There was no significant antenatal history of any infection, drug intake, hypertension or diabetes mellitus or radiation exposure to mother. The marriage was non-consanguineous. Ultrasoundography revealed a single live intrauterine foetus of 16 weeks with no congenital anomalies. She was advised to continue pregnancy. On 29th week she was admitted with leaking per vaginum (P/V) and pain abdomen. Per abdominal examination revealed uterus to be 28 weeks, foetal heart rate was 140/minute. In P/V examination, cervix was found to be 3 cm dilated with absence of membrane, hand prolapse and a mass was felt by the side of hand. Lower segment caesarean section was done and a dead foetus with multiple congenital anomalies was delivered. The dead foetus was sent to Pathology Department for detail autopsy examination.

External examination of the foetus-Weight of the foetus was 2.6 kg, the crown rump length was 23cm, crown heel length-37cm, head circumference-31cm, chest circumference-22cm and foot length was 6cm. Ear cartilage, hair, nails were well developed, palmar creases absent, few plantar creases were present. The placenta and umbilical cord were found to be normal except for single umbilical artery in cord.

Congenital malformations
- A defect in the lower anterior abdominal wall (omphalocele) of 20 x 15 cm size was present containing liver, spleen, stomach, duodenum, coils of small and large intestine and one side kidney, covered by a thick greyish white membrane (Fig.1).
- External genitalia was absent (Fig. 2A).
- Imperforate anus - In the perineal region there was no anal opening (Fig. 2A).
- Right foot showed talipesequinovalgus (Fig. 2B).
- Absent appendix. Absent gonads & Single kidney (Fig. 2C).
- Exstrophy of meconium stained bladder cloaca (Fig. 2D).
- Lumbo-sacral meningocele of 6.5cm diameter was present containing 16ml straw colored fluid (Fig. 3).
- Fetal spine was showing scoliosis (Fig. 4).

3. Discussion

OEIS complex is one of the most severe congenital abnormalities compatible with intrauterine life. It is a non-random association of 4 congenital anomalies like omphalocele, exstrophy of the cloaca, imperforate anus, and spinal abnormalities. It results from improper closure of ventral abdominal wall due to failure of cephalo-caudal and lateral foldings with associated defects of cloaca and urorectal septum. It usually occurs during early blastogenesis around 4th week of development..
OEIS complex is difficult to diagnose prenatally and all the abnormalities in the foetus may not be clear until the postnatal examination is completed. The major criteria for the prenatal diagnosis are non visualisation of foetal bladder, infra-umbilical anterior abdominal wall defect, omphalocele and myelomeningocele (MMC). The minor criteria are– lower extremity abnormalities (club foot), renal anomalies, ascites, widened pubic arches, small bowel atresia and abdominal musculature deficiency. There were many associated abnormalities in the present case also like absence of external genitalia, gonads, appendix, imperforate anus, presence of single kidney, exstrophy of meconium stained bladder/cloaca, lumbo-sacral meningocele and limb deformity as talipes equinovalgus in right foot, There was single umbilical artery in the umbilical cord which one of the minor criteria for prenatal diagnosis. Usually the cases of OEIS complex are sporadic. But according to many authors, the aetiology of OEIS complex was thought to be multifactorial. Higher incidence of OEIS in monozygotic twins than in dizygotic twins suggests a genetic contribution to the occurrence of these defects. Besides chromosomal anomalies, fungal toxin ochratoxin A, the antiparasitic drug suramin and trypan blue have been variably associated with cloacalexstrophy in chicks. In humans, maternal exposure to diphenylhydantoin, diazepam, valproeic acid, maternal obesity, diabetes mellitus and multiple uterine fibroids have been found frequently with this complex. Some cases of in vitro fertilization also suffer from this syndrome.

The present case did not have any of the risk factors and the 4 living siblings did not suffer from it. So this makes our case as one of the sporadic occurrences of OEIS complex.

Since almost all the systems in the body are variably involved by OEIS complex. The clinical presentation varies from case to case. So a multidisciplinary approach by a team including neonatologists, pediatric surgeons, pediatric urologists, pediatric neurosurgeons and pediatricorthopedic surgeons is necessary for the proper management of such cases. Immediate management is directed to the medical stabilization of the infant. Evaluation and appropriate surgical management of associated malformations should be done in subsequent frequent steps.

The birth of a congenitally deformed child imparts strong social stigma over the parents. Compatibility with life is nil or minimal with OEIS complex .Since no particular etiological factor has been found, prenatal diagnosis relies on sonographic findings. Hence, regular sonographic evaluation in the pregnant women mayhelp for early diagnosis and necessarytherapeutic intervention. This case is reported due to its rarity, associated anomalies and also for counselling of parents about future conceptions. Its future prospect is that antenatal diagnosis of this condition will help in reducing the incidence of this highly lethal condition.

References

[2] Carey JC. Exstrophy of the cloaca and the OEIS complex: One and the same(Letter)

Figure Legends

Fig. 1- Foetus with large omphalocele
Fig. 2A- Imperforate anus and absence of external genitalia
Fig. 2B- Talipes equines valgus in right foot
Fig. 2C-Internal abdominal organs with single kidney
Fig. 2D- Meconium stained bladder/cloaca
Fig. 3- Meningocele containing fluid
Fig. 4- Scoliosis of vertebral column
Figure 1: Foetus with large Omphalocele

Figure 2a, 2b, 2c, 2d: Foetus with large Omphalocele

Figure 3: Meningocele containing fluid

Figure 4: Scoliosis of vertebral column