

CASE REPORT

OMPHALOCELE, EXTROPHY OF CLOACA, IMPERFORATE ANUS AND SPINE ABNORMALITIES–OEIS COMPLEX: A RARE CASE REPORT

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HOW TO CITE THIS ARTICLE:

Uram Aruna Jyothi, B. A. Ramakrishna, K. Vandana. "Omphalocele, Extrophy of Cloaca, Imperforate Anus and Spine Abnormalities–OEIS Complex: A Rare Case Report". Journal of Evolution of Medical and Dental Sciences 2015; Vol. 4, Issue 51, June 25; Page: 8954-8957, DOI: 10.14260/jemds/2015/1299

ABSTRACT: OEIS complex is a rare defect with estimated incidence of 1 in 200,000 live births. These defects occur due to improper closure of ventral abdominal wall and constitute a spectrum ranging from simple epispadias to complex association of OEIS. Most cases are sporadic with no obvious etiology. A third gravida with previous normal obstetric history delivered a dead fetus at 18wks of gestation. The anomalies presented are grouped under OEIS complex.

KEYWORDS: Omphalocele, Extrophy of Cloaca, Spine Abnormalities.

INTRODUCTION: CASE HISTORY: A third gravida of 30yrs age with previous normal obstetric history delivered a dead fetus with multiple congenital anomalies at 20wks of gestation. [Fig. 1] The mother had a non-consanguineous marriage and she did not have any antenatal history of drug intake or infection. USG–scan during antenatal period revealed the fetus to have omphalocele with polyhydramnios and kyphosis of lumbar spine. Birth weight a fetus was 750gms. On external examination head circumference was 20 cm crown rump length was 11 and crown heel length was 14cm. The sex was indeterminate as the fetus had ambiguous genitalia with prominent rugated labiosacral folds and no apparent genital tubercle. The baby had hydrocephalus, kyphoscoliosis of lumbar spine. The thorax showed no abnormality. Lower anterior abdominal wall was found to be defective (Omphalocele) 14x11cm and containing liver stomach, spleen and loops of intestine. [Fig. 2] The omphalocele presented with ruptured sac. Further examination of eviscerated organs revealed that liver is so enlarged that it is almost found to be in the midline. The situs was normal with stomach, spleen, cecum with appendix and colon to the left. The colon presented with foci of atresia and stenosis. Both kidneys and adrenals were normal. [Fig. 3]

The diaphragm was normal. On thoracotomy lungs and thymus were found to be normal. External genitalia were not found. In perianal region there was no anal opening - imperforate anus. Both lower limbs were rotated to one side. fetal spine showed kyphoscoliosis and Extrophy of bladder cloaca mucosa is seen. There was a lumbosacral meningocele and there is a attached umbilical cord of size 12cm and shows 2 arteries and 1 vein.

DISCUSSION: The term OEIS complex which consists of omphalocele, extrophy of bladder, imperforate anus and spine abnormalities was first coined by Carey et al in 1978.¹ The incidence of OEIS complex is 1 in 200,000 to 1 in 400,000 pregnancies.¹ Most cases are sporadic with no obvious etiology. This is a rare complex disorder and very few instances of recurrence of anomalies in this cluster have been reported. In addition to the 4 classic malformations, a strong association with spina bifida and intersex was noticed.^{2,3} In a study performed by Bohria et al,⁴ it has been suggested and had pointed out that bladder extrophy and cloacal extrophy are two distinct clinical entities.⁵ It is thought that this OEIS complex results from a single defect of early blastogenesis or defect of a caudal mesodermal migration during primitive streak period that later contributes to the formation of infra umbilical mesenchyme, cloacal septum and vertebrae. Ureters could be traced up to the exposed

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cloacal mucosa. The gonads could not be traced and there were no mullerian duct, uterus or fallopian tubes.

CHROMOSOMAL ASSOCIATION: Though most causes of OEIS are sporadic with no obvious etiology, it has been suggested that it is associated with some chromosomal aberrations.² The sporadic occurrence of OEIS complex suggest both environmental and genetic factor may play a role in its etiology. Grey et al and Keppler et al⁶ had stated that cloacal extrophy is associated with maternal exposure to diphenyl hydantoin and valproic acid and cigarette smoking. Cloacal extrophy has been reported in patients with chromosomal anomalies including 3q12.2 -3q13.2 deletion⁷ and trisomy 18. Single gene defects may also head to OEIS complex altogether definitive evidence is lacking.¹

It has been reported that homologue genes such as H2xB9 and retenoic acid may play a role in OEIS complex.⁵ There is a single first of reported case of OEIS complex associate with chromosome 1p deletion.^{7,8}

OEIS complex is caused by recessive mutations of gene located in 1p36 region and deletion involved a mutation located in the exact homologue.^{1,6,7}

EMBRYOLOGICAL BASIS: There are different explanations given for the occurrence of extrophy of cloaca.⁴ A study done by marshall and mavericke⁹ stated that the overdeveloped cloacal membrane is the basic defect and this presents migration of mesoderm between ectodermal and endodermal layers. Rupture of this unduly damage cloacal membrane before the descent of urorectal septum results in extra cloaca being exposed to exterior.^{2,9} But another study done by beaudon S. et al suggested that failure of volation of pelvic bone primordia results in failure of final closure of ventral abdominal wall resulting in varying degree of extrophy.^{5,10} Ultimately it appears that, there is a defect in the gastrulation in the caudal part of embryo, caudal displacement of phallus, and defect in the urorectal septum all of which leads to extrophy of cloaca.^{7,11,12} Lack of mesoderm in the infraumbilical abdominal wall results in omphalocele.

Associated Anomalies:

Different anomalies can be associated with oeis complex.

Cardiac anomalies, renal anomalies, increased nuchal translucency markedly.

Elevated serum levels of alfa fetoprotein are always present. But most cases of oeis.

Eases are diagnosed only at autopsy after interruption of pregnancy.

CONCLUSION: Accurate prenatal diagnosis of oeis complex associated with malformations is important for detailed counseling of the family, appropriate perinatal management by the obstetrician, pediatric, neonatologist and especially pathologist.

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Fig. 1



Fig. 2



Fig. 3



Fig. 4

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Fig. 5



Fig. 6



Fig. 7



Fig. 8

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FINANCIAL OR OTHER

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Date of Submission: 06/06/2015.

Date of Peer Review: 07/06/2015.

Date of Acceptance: 22/06/2015.

Date of Publishing: 25/06/2015.