

## OEIS COMPLEX: A rare Case Report with Review of Literature.

B. Mallikarjunappa, Abhishek Ghosh

Department of Radiodiagnosis, Sri Adichunchanagiri Institute of Medical Sciences,  
B.G. Nagara, Nagamangala Taluk, Mandya District, Karnataka, India.

**Abstract:** OEIS Complex is rare congenital anomaly. The spinal deformity was Caudal Regression. We present here an antenatal case presented to us with 9 months amenorrhea, detected on ultrasound; severe form of disease is not compatible with life.

### INTRODUCTION

Omphalocele-Exstrophy-Imperforate anus-Spinal defects (OEIS) are a rare complex. OEIS represents the most severe form of Epispadias-exstrophy sequence ranging from phallic separation with epispadias, pubic diastasis, exstrophy of the bladder (isolated), cloacalexstrophy to OEIS<sup>1,2</sup>. Exstrophy of the cloaca is a well-known malformation that includes the persistence and the exstrophy of a cloaca that receives ureters, ileum and a rudimentary hindgut. This anomaly is associated with failure of fusion of the genital tubercles and pubis rami, incomplete development of the lumbo-sacral vertebrae with spinal dysraphism, imperforate anus, cryptorchidism, epispadias, anomalies of mullerian duct in females and urinary tract anomalies. Then OEIS acronym is used<sup>3</sup>. Synonyms includes Cloacal dystrophy; Vesicointestinal fissure; Splanchnicaexstrophia; Exstrophy-epispadias sequence

### CASE REPORT

A 20 year old female G1P1 was referred to our department for term scanning. Ultrasound findings included: (1.) absence of caudal vertebrae, with deformed lower limbs, very short and hypoplastic (Figure 1,2); (2.) There was a soft tissue connecting ipsilateral thighs and legs (pterygium) on both the sides. (Figure 2); (3.) There was omphalocele and bi-lateral talipes.(Figure 3); (4.) There was also imperforate anus (Figure 2); (5.) Upper extremities were normal; (6.) There was single umbilical artery.

Antenatal MRI was done which confirmed the diagnosis. Fetus was delivered by vaginal delivery and it was a live birth. Heart beat was present for 5 minutes. The weight of the fetus was 2.5 kg. It was not compatible with life.

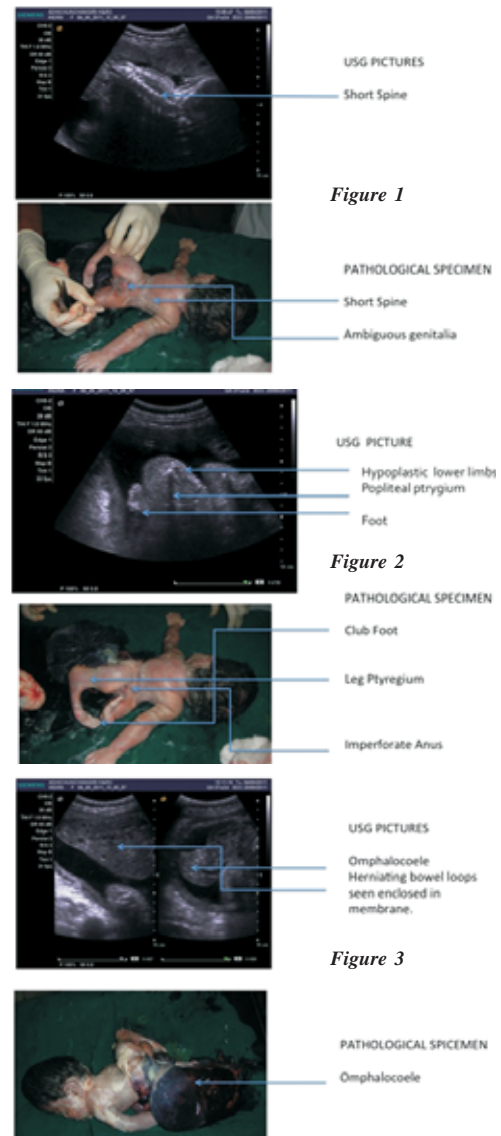
### DISCUSSION

The fetal OEIS is a defect that affects the midline of the lower inferior body. OEIS occurs with an incidence of 1:250,000 live births. The occurrence of exstrophy of the bladder appears to be more common (1:30 000 to 1:40 000) than exstrophy of cloaca (1:200,000 to 250,000) or OEIS (1:200,000 to 1:400,000) pregnancies (6). The incidence of OEIS is probably higher because many cases are diagnosed incorrectly as omphalocele, which is the most prominent component of this malformation complex<sup>7</sup>.

OEIS etiology is unknown, but several associations have been suggested:

(a) Teratogenic exposition: diazepam, diphenylhydantoin<sup>3</sup>; (b) Genetic factors: possibly associated (47 XXX; T18)<sup>8</sup>; (c) Single defects in blastogenesis and mutations in Homeobox genes (such as HLXB9). HLXB9 has also been described in association with the Currarino triad (associated sacral agenesis, anal defect including imperforate anus and anal stenosis, presacral teratoma and spinal cord tethering)<sup>3</sup>; (d) Multiple pregnancies: Higher incidence of OEIS in monozygotic twins suggests a possible genetic contribution to the occurrence of this multisystem defect<sup>2</sup>. Indeed, Lee et al describe a case of OEIS complex in monozygotic twins. This concordance of monozygotic twins for the defects may support the theory that early malformation complexes as OEIS and monozygotic twinning are manifestations of the same discordance of early blastogenesis<sup>7</sup>; (e) Sporadic familial occurrence: the most frequently etiology; (f) OEIS is considered to be a defect in blastogenesis, beginning in the first four weeks of human development. According to different authors, OEIS has probably a heterogeneous etiology and may result from a single localized defect in early caudal mesoderm at approximately 29 days of development and it is thought to lead to one of three defects<sup>9</sup>; (g)

**Correspondence:** Dr.Mallikarjunappa B., Department of Radiodiagnosis, Adichunchanagiri Institute of Medical Sciences, B.G. Nagara, Nagamangala, Mandya District, Karnataka-571448, India. e-mail: drmagarajbt@gmail.com



Failure of cloacaseptation: resulting in a common cloaca. (Figure 1, 2); (g) Breakdown of the cloacal membrane: resulting in exstrophy and omphalocele; (h) Incomplete vertebral fusion: resulting in open neural defect (Spina bifida). Any abnormality in the formation of uro-rectal septum results in the failure of urogenital sinus to separate from the rectum. The mesodermal proliferation forming the infra-umbilical abdominal wall and genital tubercle fail to develop. Other associated anomalies are hypoplastic chest, diaphragmatic hernia, meningocele, two vessel cord, vertebral anomalies. **Sonographic findings:** Austin et al described major criteria for the prenatal diagnosis of OEIS (non visualization of the fetal bladder, infra-umbilical abdominal wall defect, omphalocele, myelomeningocele) and minor criteria (lower extremities malformations, renal anomalies, ascites, widened pubic

arches, narrow thorax, hydrocephalus, single umbilical artery)<sup>6</sup>.

According to the literature, sonographic findings of OEIS complex have been documented, but only few cases of prenatal diagnosis have been reported. Nevertheless, nowadays, the **prenatal diagnosis** of OEIS is possible by the identification of:

- A midline infra-umbilical defect with an irregular mass: in the inferior abdominal wall or cystic anterior wall structure (persistent cloacal membrane) or with omphalocele. Prenatal detection of anterior abdominal wall defect has significantly improved over last few years. It is among the more definitive diagnosis that can be made in a routine obstetric ultrasound examination. The Standard views required for prenatal ultrasound examinations, as per the guidelines of American College of Radiology, are the demonstrations of the umbilical cord insertion over foetal abdomen and the integrity of the anterior abdominal wall<sup>12</sup>.
- Absence of the bladder between the two umbilical arteries.
- Lumbo-sacral myelomeningocele: always seen. Common spinal defect include hemivertebrae, sacral anomalies and either tethered cord and meningocele. But it is important to know that Källén et al wrote that the spinal defect may occur more cranially and are not restricted to the lumbo-sacral region<sup>8</sup>. Our case had caudal regression, there was abrupt disruption of vertebrae. The pathognomic feature is absence of vertebrae in axial section of the fetal abdomen.
- Anomalies of the inferior limbs are possible but club feet, limb duplication or amputations are generally not seen with OEIS<sup>10</sup>. Our case was a rare entity having club foot. (Figure 2)
- Wide pubic arch is classically present with symphysis pubis diastasis and congenital hip dislocation.
- Single umbilical artery: It is a frequent associated sign<sup>11</sup>.
- Genital anomalies. Classically, the sex determination is often not possible. Other uro-genital anomalies are possible, including genital duplication. (Figure 1)
- Anal atresia. (Figure 2)
- Omphalocele. (Figure 3)

Majority of authors consider OEIS as a distinct syndrome, but there is a discussion if the exstrophy of the bladder sequence, exstrophy of the cloaca sequence or urorectal septum malformation sequence should be referred to distinct clinical entities.

**Associated Anomalies:** Different anomalies can be associated with OEIS complex:

- Cardiac anomalies: cardiac defects have been described with exstrophy of the cloaca alone (as atrial and ventricular defect).
- Renal anomalies
- Increased nuchal translucency: Only one case associated with OEIS was described by Schem et al<sup>2</sup>. The most likely cause of increased nuchal translucency is probably vascular or hemodynamic, but the mechanism remains unclear. Nevertheless, there is no evidence for jugular lymphatic obstruction sequence in OEIS.

Markedly elevated serum levels of alpha fetoprotein is always present. But most cases of OEIS complex are diagnosed only at autopsy after interruption of pregnancy.

**Prognosis:** The prognosis of infants with OEIS complex is variable, depending on the severity of the structural defects. Survival will depend on the extension of the cloacalexstrophy and the neural tube defect. In less severe forms, good outcome with corrective surgery is possible but very uncommon. Cloacalexstrophy is lethal due to obstruction of the urinary tract and association with renal and pulmonary complications. So early prenatal diagnosis of OEIS complex is required to give parents the option to terminate the pregnancy. And it is also helpful to plan the appropriate perinatal management. In cases where parents decided to continue the pregnancy, serial scans are necessary to evaluate the progress of the ventriculomegaly, which can be associated with OEIS. But, we think that cesarean section should be performed in cases of OEIS complex to avoid dystocia and trauma.

**Management:** It seems essential to perform an early scan in the next pregnancy, because an accurate prenatal diagnosis of OEIS complex associated with malformations, is important for the detailed counseling of the family (interruption of pregnancy) and appropriate perinatal management by the obstetricians, pediatric surgeons, neurosurgeons, neonatologist, but especially anatomic-pathologist.

## CONCLUSION

USG is the imaging modality of choice to diagnose OEIS Complex in the 2<sup>nd</sup> trimester. MRI can be useful back up to the USG findings.

## REFERENCES

- 1 Chih-Ping chen, Shin-Lin Shih, Fen-fen Liu, Sheau-Wen Jan, Cherng-Jye Jeng, Chung-Chi lan-Exstrophy-Imperforate anus-Spinal defects (OEIS) associated with large meningocele and severe limb defects. *Am J Perinatol* 1997 ; 14 : 275-80.
- 2 Noack F, Sayk F, Gembruch U.-Omphalocele-exstrophy-imperforate anus-spinal defects complex in dizygotic twins. *Fetal Diagn Therapy* 2005 ; 20 : 346-8.
- 3 Shanske A.L., Pande S., Aref K., Vega-Ric C., Brion L., Reznik S., Timor-Trisch I.E.- Omphalocele-exstrophy-imperforate anus-spinal defects (OEIS) in triplet pregnancy after IVF and CVS. *Birth defects research (part A)* 2003 ; 67 : 467-71
- 4 Güz B.A., Sherer D.M., Atkin J., Venanzi M., Ahlborn L., Cestone L.- Firt-trimester prenatal sonographic findings associated with OEIS
- 5 Smith N.M., Chambers H.M., Furness M.E., Haan E.A.- the OEIS complex : recurrence in sibs. *J med Genet* 1992 ; 29 : 730-2. complex ; a case and review of the literature. *Am J Perinatol* 1998 ; 15 : 15-7.
- 6 Austin P.F., Homsy Y.L., Gearhart J.P., Porter K., Guidi C., madsen K., maizels M.- The prenatal diagnosis of cloacalexstrophy.
- 7 Lee D.H., Cottrell J.R., Sanders R.C., Meyers C.M., Wulfsberg E.A., Sun C.C.-J- OEIS complex (Omphalocele-Exstrophy-Imperforate anus-Spinal defects) in Monozygotic twins. *Am J med genetics* 1999 ; 84 : 29-33.
- 8 Källén K., Castilla E.E., Robert E., Mastroiacovo P., Käille P.- OEIS complex a population study. *Am J med Genet* 2000 ; 92 : 62-8.
- 9 Haldar A., Sharma A.K., Phadke S.R., Jain A., Agarwal S.S.- OEIS Complex with cranio-facial anomalies- defect or blastogenesis. *Am J Med Genetics* 1994 ; 53 : 21-3.
- 11 Joung-Liang Wu ; Kung-Hong Fang ; Guang-Perng Yeh ; pan-Hsin Chou ; Charles Tsung-Che Hsieh- Using color dopplersonography to identify the vesical umbilical artery. *J Ultrasound Med* 2004 ; 23 : 1211-5.
- 12 HR Shah, TC Patwa, AB Tannk, JB Pandya, et al. *Gynecology and obstetrics ; A Case of fetus having combined features of LBWC + OEIS complex. Indian Journal of Radiology and Imaging.* 2005;15(1):85-8.

## JIMSA TRAVEL GRANT - 2015

The International Medical Sciences Academy is pleased to announce JIMSA Travel Grant to Research Scholars for presentation at IMSA's annual conference IMSACON every alternate year for travel within the country. The Award shall be in conformity with the following guidelines:

- 1) No. of Grants – Two (2);
- 2) Travel Grant not exceeding Rs. 8000/- per awardee, to cover rail travel expenses whichever is less.
- 3) Original Research Work by a young researcher (age < 45 years) for presentation at IMSACON every alternate year for travel within the country.
- 4) Research Work should clearly project the objectives, selection of material, methodology adopted, result analysis with statistics, discussion and conclusions. A summary in 350 words highlighting why the paper should be considered for the award, must be enclosed.
- 5) In case the applicant is in Government job, he/she should enclose a letter from the Head of Department / Institution, certifying that he/she is not being supported by any other agency.
- 6) The applicant should send to Editor JIMSA, abstract of article alongwith the copy of acceptance letter from Chairman, scientific Committee, of IMSACON 2013 for consideration of the JIMSA AWARDS COMMITTEE. The selected candidates will be given the grant at the venue of IMSACON subject to the production of participation certificate.

The selected article (Full Text) will be considered for priority publication in JIMSA only after the proper peer review by the referee.

Dr. P. D. Gulati  
Editor, JIMSA

The above guidelines are in adherence to and in conformity with the decisions in the meetings of Central Executive Committee and in the meeting of Board of Trustees of International Medical Sciences Academy, World Headquarters, New Delhi held on January 26, 2010.