Journal of Clinical and Biomedical Sciences

Journal homepage: www.jcbsonline.ac.in

Case Report

OEIS Complex–Diagnosis By Fetal Magnetic Resonance Imaging and Confirmation By Autopsy: A Case Report.

G Sarat Chandra[1]*, R Hemalatha[2], B Umamaheshwari[1]

1. Department of Neonatology, Sri Ramachandra Medical College & Research Institute, Chennai .

2. Department of OBG, Consultant Sonologist, Sri Ramachandra Medical College & Research Institute, Chennai.

Received: 16th October-2014 Accepted: 6th December-2014 Published: 31st-December2014

Abstract

A 26 year old was referred to us in view of an abnormal anomaly scan. Ultrasonograph showed omphanocoel and extrophy of cloaca, complete spinal defects couldn't be commented upon. Fetal MRI aided in the diagnosis and the final diagnosis was confirmed by autopsy. Our case report suggest that fetal MRI is a useful tool for prenatal diagnosis and helps in prenatal counseling or planning of postnatal early treatment strategy.

Key words: OEIS, Fetal MRI, Fetal autopsy, congenital defects

Introduction

Omphalocele– extrophy of cloaca - Imperforate anus– spinal defects (OEIS) complex, with its unknown etiology is a rare disorder, which is incompatible with life. Term was coined by Carey et al, but the first description of the case was published by Litter in 1709. We present a rare case that was diagnosed antenatally by second trimester ultrasound scan, confirmed by a fetal MRI and detailed autopsy findings.

Case History

A 26 year old was referred to us in view of an abnormal anomaly scan. A three generation detailed pedigree chart did not reveal any abnormality. With no consanguinity, her first child is a 6 year old healthy male, second is a medical abortion for personal reasons and third was the current pregnancy. Second trimester screen showed omphanocoel and extrophy of cloaca, but the length of spinal defects couldn't be commented upon. (Fig.1) Hence a fetal MRI was asked for, which showed a 2cm defect in the inferior aspect of the anterior abdominal wall with herniation of bowel loops and bladder. Deficiency of sacrum with normal thorac-lumbar and cervical spine was also noted clearly. (Fig. 2) Hence, the diagnosis was made based on the constitutional features of OEIS complex.

Parents were given a non-directional counselling regarding poor prognosis, quality of life and morbidity. They opted for medical termination of pregnancy. They were also counselled regarding karyotyping for 1p36 deletion and autopsy for the fetus. MTP was carried out and the fetus was sent for autopsy. Karyotyping was planned but couldn't be done due to logistic reasons.

Autopsy (Fig 3) showed that the fetus had a prominent occipital region, umbilical cord was pushed to right and a portion of intestines and the whole bladder were protruding through abdominal wall with presence of gonads on both sides. No external genital organs were seen. Imperforate anus and bilateral congenital talipes equinovarus were also present. Right kidney is in the lower iliac fossa and right adrenal was flat. Autopsy confirmed all the constitutional features of OEIS complex.

*Corresponding Author

Dr. G. Sarat Chandra, Department of Neonatology, SRMC, Chennai, India.

E mail : saratgullapalli@gmail.com.

Quick access Code

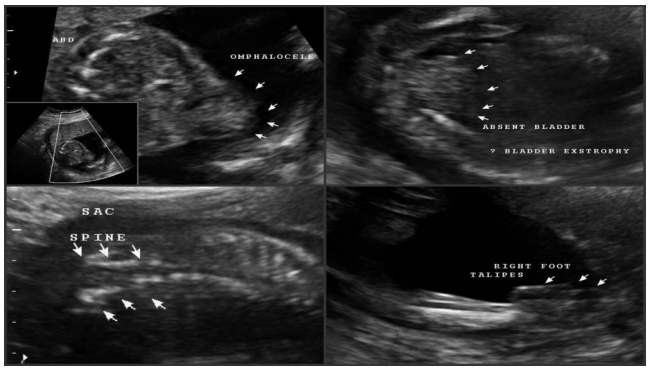

Figure 1: OEIS findings on sonography.

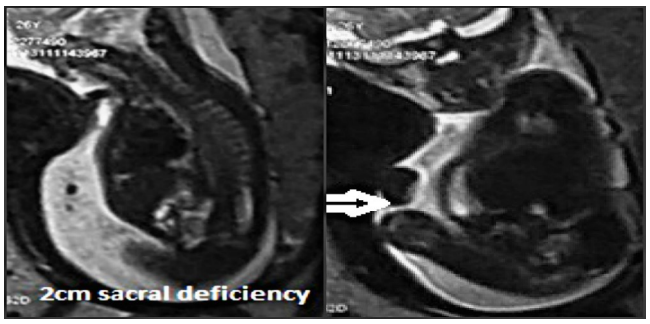

Figure 2: Fetal MRI showing sacrum deficiency, extrophy of bladder and herniation of bowel (white arrow).

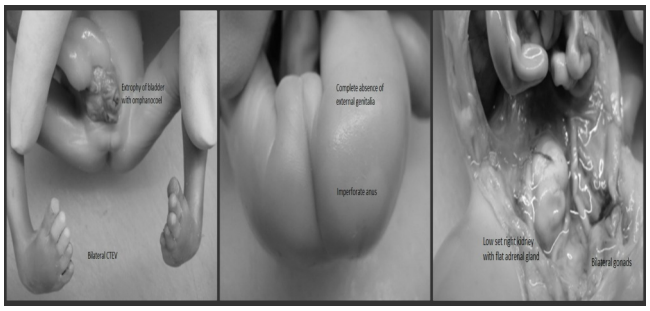

Figure.3 Fetal autopsy

Discussion

OEIS complex, an exceedingly rare defect which comprises a combination of omphalocele, extsophy of the cloaca, imperforate anus, and spinal defects and is reported at a prevalence rate of 1:100,000 live births with a male: female sex ratio of 2:1.[1] With no obvious etiology, defects were hypothesized to occur in the early blastogenesis or in the mesodermal migration during the primitive streak period. In 35–65% of the cases, cloacal extrophy is associated with the other three classical malformations (omphalocele, imperforate anus, and spine abnormalities).[1] It has a variable association with pelvic and genital abnormalities, limb abnormalities, and spina bifida, which has been described as lumbar in 72%of the cases, sacral in 14%, and thoracic in 14%.[2]

Prenatal diagnosis of OEIS complex depends on the ultrasound criteria that include an infra-umbilical abdominal wall defect with a protruding mass, absent bladder, and spinal defects.[1] MRI, an operator independent diagnostic tool can prove valuable in confirming the diagnosis.

The prognosis is variable and depends on the severity of the structural defects especially on the extension of the cloacal exstrophy and the neural tube defect. Cloacal exstrophy is lethal due to obstruction of the urinary tract and association with renal and pulmonary complications. Hence, early prenatal diagnosis is required to plan an appropriate perinatal management. [3] El-Hattab et al reported a classic malformation, in addition to renal, genital, pelvic and limb deformities with a chromosomal microarray analysis demonstrating a terminal deletion of chromosome 1p.[5] But a comprehensive genetic analysis study concludes that OEIS is a complex disorder involving a combination of genetic and environmental predispositions and is unlikely to be caused by a recurrent chromosomal aberration.[5]

References

1. Keppler-Noreuil KM. 2001. OEIS complex (omphalocele-exstrophy-imperforate anus-spinal defects): A review of 14 cases. Am J Med Genet 99:271–279.
2. Bohring A. OEIS complex. VATER, and the ongoing difficulties in terminology and delineation (Letter). Am J Med Genet 2002;107:72-76
3. Howell C, Caldamone A, Snyder H, Snyder H, Ziegler M, Duckett J. 1983. Optimal management of cloacal exstrophy. J Pediatr Surg 18:365–369.
4. El-Hattab AW, Skorupski JC, Hsieh MH, Breman AM, Patel A, Cheung SW, Craigen WJ. 2009. OEIS complex associated with chromosome 1p36 deletion: A case report and review. Am J Med Genet Part A 152A:504–511.
5. Vlangos C, Siuniak A, Ackley T, van Bokhoven H, Veltman J, Iyer R et al. Comprehensive genetic analysis of OEIS complex reveals no evidence for a recurrent microdeletion or duplication. Am J Med Genet Part A. 2010;155(1):38-49.

How to cite this article: G Sarat Chandra, R Hemalatha, B Umamaheshwari. OEIS Complex–Diagnosis By Fetal Magnetic Resonance Imaging and Confirmation By Autopsy: A Case Report. J Clin Biomed Sci 2014; 4(4):365-66.

Conflict of interest: The authors claim to have no conflict of interests in the context of this work.