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Case Report

Prenatal Diagnosis and Postnatal View Of Omphalocele-Exstrophy Vesicalis-Imperforate Anus-Spinal Defects Complex

Omfalosel-Ekstrophy Vesicalis-Imperfore Anus-Spinal Defekts Kompleksinin Prenatal Tanısı ve Postnatal Görünümü

Erdal Şeker*, Hasan Süt, Coşkun Ümit, Mustafa Koçer, Acar Koç

Department of Obstetrics and Gynecology, Ankara University School of Medicine, Ankara, Turkey

Abstract

The omphalocele-exstrophy vesicalis-imperforated anus-spinal defects (OEIS) complex is a rare variant of the bladder exstrophy epispadias complex and multisystem anomalies involving the gastrointestinal, genitourinary, and skeletal systems. The number of patients who have OEIS complex and do not terminate their pregnancy is small. We present the prenatal and postnatal radiological images of a patient diagnosed with OEIS at the twentieth gestational week.

Keywords: Omphalocele-exstrophy vesicalis-imperforate anus-spinal defects (OEIS), prenatal diagnosis, fetal anomaly

Öz

Omfalosel-ekstrofi vesicalis-imperfore anüs-spinal defektler (OEIS) kompleksi, mesane ekstrofi epispadias kompleksinin nadir bir varyantıdır. OEIS kompleksi, gastrointestinal, genitoüriner ve iskelet sistemlerini içeren bir grup multisistem anomalisidir. OEIS kompleksine sahip olup gebeliğini sonlandırmayan hasta sayısı azdır. Yirminci gebelik haftasında OEIS tanısı konan bir hastanın doğum öncesi ve doğum sonrası radyolojik görüntüsünü sunuyoruz.

Anahtar kelimeler: Omfalosel-ekstrofi vesicalis-imperforate anüs-spinal defektler (OEIS), prenatal tanı, fetal anomali

Corresponding Author *: Erdal Şeker, Department of Obstetrics and Gynecology, Ankara University School of Medicine, Balkiraz, 21, Tıp Fakültesi Cad., 06620 Mamak Ankara, Turkey
e-mail: erdalseker84@gmail.com
ORCID: 0000-0001-9818-0414
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1. Introduction

The omphalocele-exstrophy vesicalis-imperforate anus-spinal defects (OEIS) complex is a rare variant of the bladder exstrophy epispadias complex (1). The bladder exstrophy epispadias complex represents a spectrum with severity depending on the time of disruption ranging from isolated epispadias over classic bladder exstrophy to the most severe form, cloacal exstrophy. The incidence of the OEIS complex is not clear. However, in some studies, it has been reported to occur at a rate of one in 200,000 live births (2). OEIS complex is a group of multisystem anomalies including gastrointestinal, genitourinary and skeletal systems.

Since some cases have neural and abdominal defects, alpha fetoprotein (AFP) is found to be high in prenatal screening tests. However, since ultrasonography (USG) is used more widely and more frequently, it can now be diagnosed with USG at an earlier stage. There is no specific gene or teratogenic agent that can cause the OEIS complex (3). Termination option was recommended for most of the cases diagnosed prenatally and most of these cases were terminated (4). The number of patients who do not want termination is low.

Some of the cases that resulted in live birth died in the early neonatal period. Respiratory problems have been detected in the early neonatal period in babies born alive. It has been suggested that the reason for this is the accompanying axial mesodermal dysplasia on the basis of lung and caudal hypoplasia (5).

2. Case report

Our patient was 31 years old. Our patient had her third pregnancy and her previous 2 children were also healthy. The couple was non-consanguineous. There were no features in her medical history and family history, either. there was no history of exposure to any known teratogens.

In the first trimester USG scan performed in our patient at the age of 12 weeks, an omphalocele sac was found in the anterior abdominal wall. Karyotype analysis was planned for the patient for possible anomalies and prognosis. Chorionic villus sampling (CVS) was performed for genetic diagnostic testing. Genetic analysis report resulted as 46, XX. Multiple fetal anomaly was found when USG was performed in the second trimester. Sonographic findings included fetal biometry consistent with the patient's last menstrual period dating, normal amniotic fluid, and normal fetal kidneys. An absent urinary bladder, an anterior abdominal wall mass inferior to the cord insertion, a lowset abdominal umbilical cord insertion were all noted, consistent with bladder exstrophy (**Figure 1**). A sac compatible with omphalocele was observed under the place where the umbilical cord entered the abdomen. USG revealed peristalsis in this pouch and it was thought that it might be the intestine. Neural tube defect was observed in the lumbosacral region and neural tissues were found to be herniated into the sac (**Figure 2**). Anal sphincter was not observed in the examination. In the left lower extremity, it was detected that the sole of the foot was in the appearance of pes equinovarus and it was observed in the same cross section as the tibia. Termination option was offered to the patient in terms of possible prognosis and anomalies. However, the family did not accept the termination proposal.

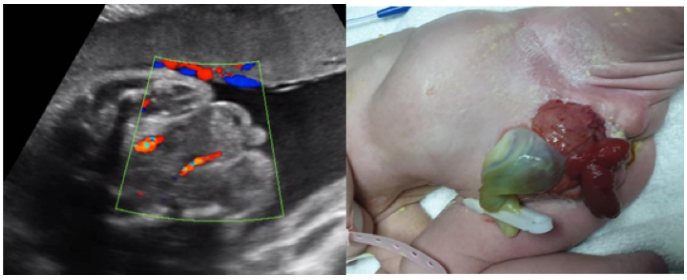

Figure 1. Exstrophy vesicalis

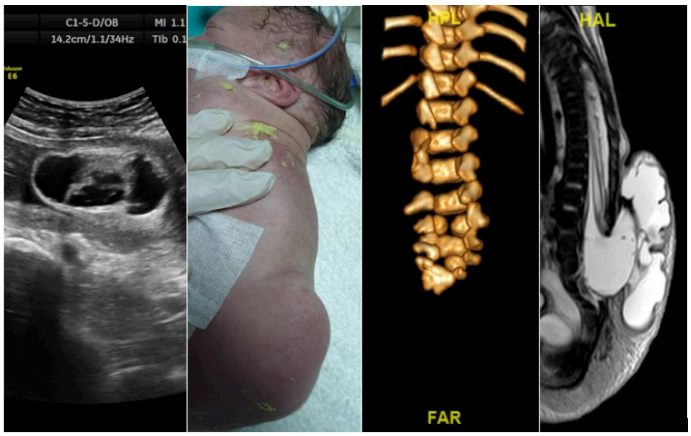

Figure 2. Spinal defects

In the follow-up of the patient, intrauterine growth retardation (IUGR) developed after the 34th week was detected. The patient, who did not have any additional problems, was taken to cesarean (CS) surgery due to his previous cesarean section when he was 38 weeks old. An ambiguous genital-looking baby was delivered at 2670 grams and 4-8 APGARs. An omphalocele sac was found in the anterior of the abdomen on the external view of the newborn. In the genital examination, genital organs were not clearly visualized due to exstrophy vesicalis. A skin-covered myelocele sac was observed in the lumbosacral region. Examination and radiographic imaging of the fetus after birth was performed. Prenatal findings were confirmed postnatally (**Figure 1, Figure 2**). The patient was admitted to the newborn intensive care unit for

further examination. The newborn underwent surgery for anal atresia, exstrophy vesicalis and meningomyelocele. Currently, the newborn is healthy and 10 months old.

3. Discussion

The OEIS complex is differentiated from other abdominal wall defects and isolated urinary system anomalies due to the coexistence of many anomalies. Usually such patients are diagnosed in the first or second trimester. As mentioned in the literature, our case presented with cystic formation in the anterior abdominal wall before cloacal membrane rupture in the first trimester (5). In these patients, not all components of the OEIS complex need to be included. In addition, renal anomalies, single umbilical artery anomalies, spleen, tracheoesophageal and duodenal anomalies accompanying the OEIS complex have also been reported in the literature (5-7).

The factors involved in its etiology are not fully known. No teratogenic or genetically specific location has been identified that can cause this disease. In the 12 disease series that Mallman presented in 2017, 10 patients were terminated. One patient died postpartum. Only one patient lived (5). Many postpartum surgeries are required for these patients. At the same time, it has high morbidity and mortality rates due to other accompanying anomalies. For these reasons, prenatal and postnatal management should be done in tertiary centers. Postnatal long-term follow-up is required due to the surgeries she will undergo.

Since the cause of the OEIS complex is not known exactly and the complication rate is high, it will remain up-to-date in the upcoming processes. In addition, elucidation of the cause will shed light on other abdominal, vertebral and urogenital anomalies.

Decleration of interest

The authors report no conflicts of interests.

Informed consent

Written consent was obtained from the family to present the case.

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