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CASE REPORT

Prenatal Diagnosis of Cloacal Exstrophy: A Case Report and Differential Diagnosis with a Simple Omphalocele

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Cloacal exstrophy is a rare congenital disorder that may lead to mortality and morbidity. Although the prenatal diagnosis of cloacal exstrophy can be made by a midtrimester ultrasound, it is difficult to differentiate it from a simple omphalocele that can be corrected completely by surgery without morbidity. We reported a case with cloacal exstrophy and reviewed previous literature on differentiating it from an omphalocele. A 33-year-old, pregnant female visited our outpatient center for prenatal care at the 22nd gestational week. The midtrimester ultrasound showed fetal anomalies including a protruding mass from umbilicus, absence of bladder, ambiguous genitalia, and bilateral renal hydronephrosis. The parents received prenatal genetic counseling and decided to continue the pregnancy. A female baby was delivered at the 37th gestational week via vaginal delivery, and cloacal exstrophy without omphalocele was diagnosed. Cloacal exstrophy is a complicated congenital disorder that should be differentiated from a simple omphalocele. Prenatal counseling and postnatal care in a tertiary medical center are important for parents and the fetus, respectively.

Conflicts of interest: The authors declare no conflicts of interest.

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Introduction

Cloacal exstrophy is a rare congenital disorder that may lead to mortality and morbidity. It is characterized by an infraumbilical abdominal wall defect, incomplete closure of the bladder with mucosa continuous with the abdominal wall, epispadias, and alterations in the pelvic bones and muscles [1]. Besides, the affected infants may also be associated with other anatomic anomalies, including gastrointestinal, genitourinary, central nervous system, and skeletal problems [2]. The incidence of cloaca exstrophy is between 1/200,000 and 1/400,000 live births [2]. The survival rate can reach to 100% with present advances in neonatal care and surgical technique [2]. Thus, early prenatal diagnosis and transfer to a tertiary medical center for further neonatal care are very important for these newborns. However, early prenatal diagnosis of a cloacal exstrophy remains a challenge because of its complicated associated anomalies [3]. In a serial study, only 25% of cases can accurately be diagnosed prenatally [3]. In this study, even those pregnancies that did not result in a live birth were taken into account; a detection rate of only 33% could be achieved [3].

Herein, we reported a case of cloacal exstrophy that mimics a simple omphalocele in the initial midtrimester ultrasound examination. Further evaluation confirmed the diagnosis of cloacal exstrophy. The literature was also reviewed to help differentiate a simple omphalocele from cloacal exstrophy.

Case Report

A 33-year-old, gravid 1, para 0, pregnant female visited our outpatient center at the 22nd gestational week. She

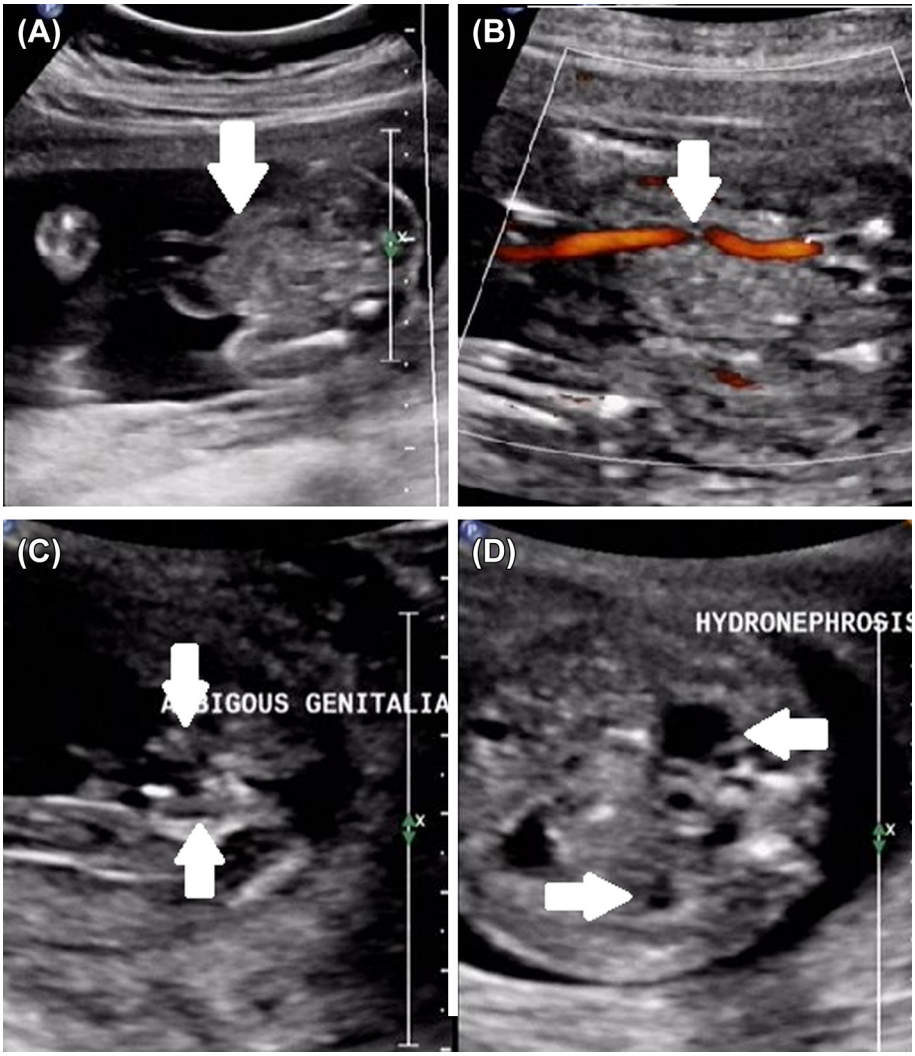

Fig. 1 Midtrimester ultrasound images of cloacal exstrophy that may mimic an omphalocele. (A) The transverse view of abdomen showing a protruding mass at the base of umbilicus (white arrow). This protruding mass was surrounded by a hyperechoic membrane with heterogeneous echoic component inside. (B) The bladder could not be identified accurately. The two umbilical arteries were traced and inserted into iliac artery directly without a full bladder between these two arteries (white arrow). (C) Between the two thighs, two small and split hyperechoic nodules, which were suspected as two hemivulva, were noted (white arrows). (D) Bilateral mild renal hydronephrosis was noted. The anterior-posterior diameters of right and left renal pelvis were measured to be 8 mm and 4 mm, respectively (white arrows).