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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.

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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

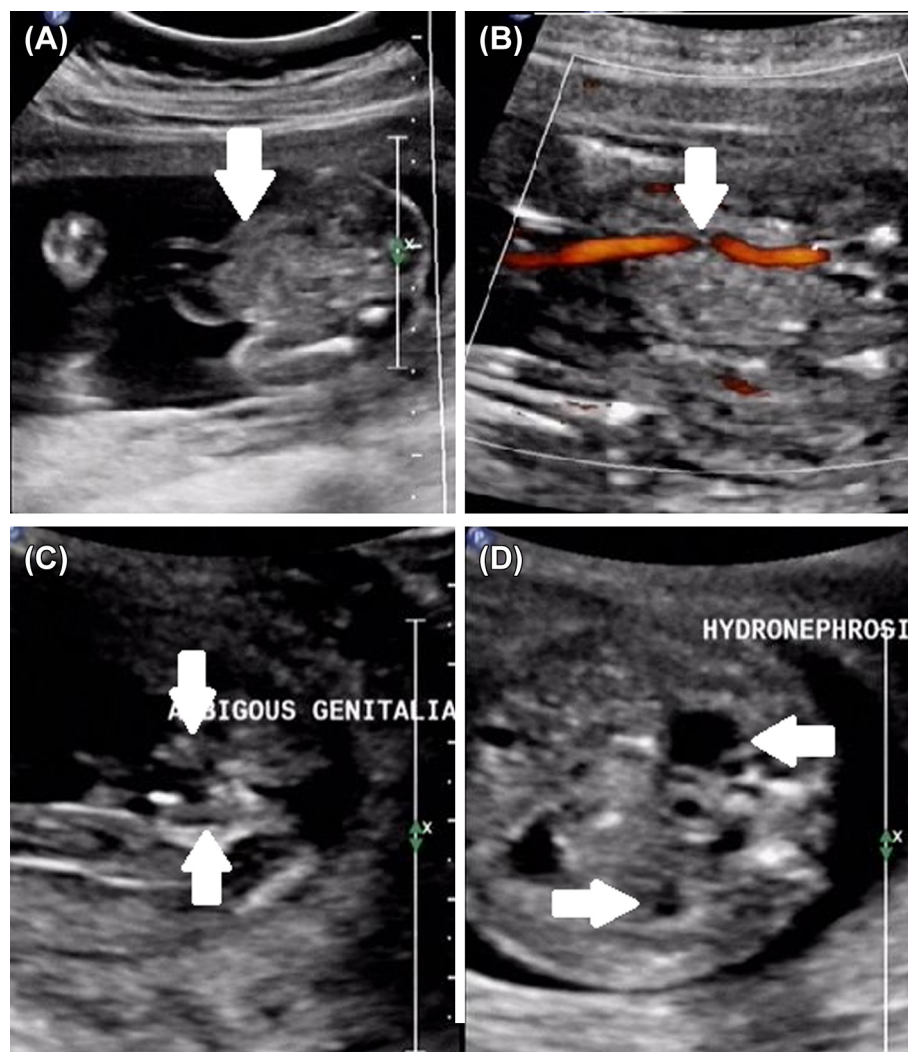


Fig. 1 Midtrimester ultrasound images of cloacal exstrophy that may mimic an omphalocele. (A) The transverse view of abdomen showing a protruding mass at the base of umbilicus (white arrow). This protruding mass was surrounded by a hyperechoic membrane with heterogeneous echoic component inside. (B) 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).

conceived naturally, and her prenatal care was uneventful prior to the 16th gestational week. However, a midtrimester ultrasound examination at the 22nd gestational week showed a protruding mass from umbilicus, and a case of omphalocele was suspected then. Thus, she was referred for further prenatal genetic counseling. Amniocentesis was performed, and the karyotyping showed 46, XX. After comprehensive prenatal counseling, the parents decided to continue the pregnancy.

In the midtrimester prenatal ultrasound examination, the transverse view of the abdomen showed a protruding mass at the base of the umbilicus (Fig. 1A). This protruding mass was surrounded by a hyperechoic membrane with heterogeneous echoic component inside. Initially, an omphalocele was highly suspected. The bladder cannot be identified clearly. The two umbilical arteries were traced and inserted into the iliac artery directly without a full bladder between these two arteries (Fig. 1B). The sex of the fetus cannot be determined by the ultrasound examination due to the ambiguous genitalia. Between the two thighs, two small and split hyperechoic nodules, which were speculated as female hemivulva, were found (Fig. 1C). The amniotic fluid index was within normal limit.

In addition, bilateral mild renal hydronephrosis was also noted. The anterior-posterior diameters of the right and left renal pelvis were measured up to 8 mm and 4 mm, respectively (Fig. 1D). No other abnormal lesions were found in the sonographic examination.

At the 37th gestational week, this patient smoothly delivered a female newborn via vaginal delivery. The female newborn weighted 2625 g, with an Apgar score of 8 at 1 minute and 9 at 5 minutes, respectively. The general appearance of the baby revealed an umbilical cord with a low insertion (Fig. 2A). However, no omphalocele was found. Beneath the insertion of the umbilical cord, two reddish protruding masses were prominent and bifid exstrophy of the bladder was suspected. Along with the defect of the pubic bone, anterior displacement of the anus with meconium attached to it was noted below the exstrophy of the bladder. A wild and split hemivulva was noted at the inner side of the bilateral thighs (Fig. 2B).

This female newborn received the first operation for bladder repair, colostomy, symphysis closure, and vaginal repair. After 4 months, she received the second operation for symphysis repair, vaginal plasty, and colon reanastomosis. Her condition was well after surgery.

Discussion

The prenatal diagnosis of cloacal exstrophy is still a great challenge for clinicians due to the lack of a reliable sign. In the study of Gearhart et al [4], a low-set umbilicus, a wide ramus pubis, diminutive genitalia, and a lower abdominal mass were the key factors for diagnosis of cloacal exstrophy [3]. However, an omphalocele may be initially suspected at the first ultrasound examination in these cases with the characteristic of a protruding abdominal mass around the umbilicus [5]. Absence of a bladder became a crucial sign for confirming the diagnosis of cloacal exstrophy [4]. Lee et al [6] reported that, apart from the absence of a bladder, a solid mass in the lower fetal abdomen with umbilical arteries running alongside the mass is helpful for the diagnosis of cloacal exstrophy. Furthermore, Goldstein et al [7] reported a case in which the urachus resembled the normal bladder. Thus, considering that nonvisualization of the bladder is one of the main findings in cases with cloacal exstrophy, special attention should be given to fetal lower abdominal cystic structures in order to differentiate them from a normally positioned fetal bladder [7]. In our case, we found a low-set umbilicus, diminutive genitalia, and a lower abdominal mass, which are compatible with the characteristics described in previous literatures [9]. However, no omphalocele was found after delivery. As the quality of life after surgery was different between a simple omphalocele and a cloacal exstrophy, an accurate prenatal diagnosis is very important for these fetuses and then the parents can be provided necessary prenatal counseling. Fetuses should be transferred to a tertiary medical center for better neonatal care [8].

In addition to the abnormality of the abdominal wall and the bladder, ambiguous genitalia were also very common

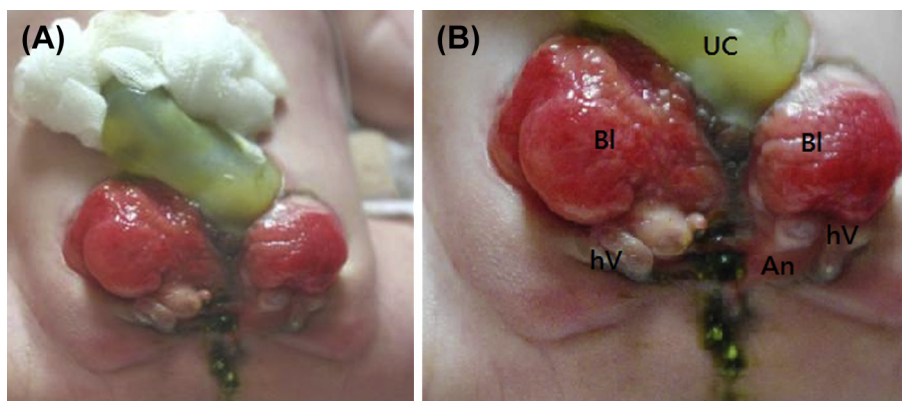


Fig. 2 (A) General appearance of the delivered newborn revealed an umbilical cord with low insertion. (B) Details of cloacal exstrophy. An = anterior displacement of anus filled with meconium, which was noted below the exstrophy of bladder; Bl = bifid exstrophy of bladder; hV = wild and split hemi-vulva at the inner side of bilateral thighs; UC = umbilical cord, which is low insertion but without omphalocele.

among these cases [2]. Complete separation or absence of the phallic or clitoral halves may be seen [2]. In prenatal ultrasound, the genitalia are difficult to identify and may appear as two little nodules, widely separated, between the bilateral fetal thighs, as in our case. Further karyotyping may be necessary for the confirmation of fetal sex. To sum up, an experienced sonographer should be aware of the diagnosis of cloacal exstrophy if the prenatal ultrasound showed an abdominal protruding mass, absence of the bladder, and ambiguous genitalia. In addition, further detailed examination is necessary.

After delivery, the newborn with cloacal exstrophy requires a surgery of the bladder and associated structural repair [1]. One of the primary goals of repair is to maintain urine continence, defined as dry intervals of ≥ 3 hours [1]. In the study of Husmann et al [9], widely reported results related to complete urinary continence and volitional voiding have been published, with urinary continence reported to occur in 7–85% of patients. Besides, a cosmetically acceptable and functional phallus can be achieved in 85% of patients [9].

With the advanced image technology, a more accurate diagnosis of cloacal exstrophy can be made by fetal magnetic resonance imaging (MRI) in a case suspected by a prenatal ultrasound examination [10]. In the study of Calvo-Garcia et al [10], fetal MRI confirmed cloacal exstrophy in all eight fetuses by demonstrating absence of a normal bladder and lack of meconium-filled rectum/colon, associated with a protuberant pelvic contour and an omphalocele. In a report by Clements et al [11], the fetal MRI revealed a bifid appearance of the bladder with bowel loops protruding through the center, which is consistent with an “elephant trunk” deformity. In their opinion, fetal MRI led to early and complete identification of the spectrum of anomalies, and facilitated verification of these findings by subsequent sonography [11].

In conclusion, an accurate prenatal diagnosis of cloacal exstrophy is very important. A low-set umbilicus, a wide ramus pubis, diminutive genitalia, a lower abdominal mass, and, most importantly, absence of a full bladder should aware the sonographer of the diagnosis of cloacal

exstrophy. In addition, fetal MRI helps in making a more accurate diagnosis. Once the diagnosis is confirmed, parents should receive prenatal counseling and have the option of terminating this pregnancy. The affected newborns can also avail of more advanced neonatal care if they can be transferred early to or delivered in a tertiary medical center.

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