Case Report

Prenatal diagnosis of cloacal anomaly in trisomy 21

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ABSTRACT

The cloacal malformation is an extremely rare non-hereditary fetal anomaly that presents as a variety of defects. It predominantly affects females with a prevalence of 1 in 50,000 births. Here, we present the case of a 27-year-old primigravida at 32 weeks of gestation showing a large midline abdominopelvic cystic mass with a septum, bilateral hydroureteronephrosis with oligohydramnios in a female fetus on antenatal ultrasonography. Suspicion of the cloacal anomaly was made which was confirmed postnatally. Postpartum analysis of the placenta revealed trisomy 21.

Key words: Cloacal anomaly, Imaging, Prenatal diagnosis, Trisomy 21, Ultrasound

The cloacal anomaly is a spectrum of congenital pelvic malformations that result from abnormal cloacal division during early embryogenesis. It is seen predominantly in females. Depending on the timing of the developmental arrest, a spectrum of abnormalities can result, ranging from urogenital sinus malformations to cloacal dysgenesis. These anomalies are rare, the prevalence being 1 in 50,000 births for the most common forms and 1 in 250,000 births for the rarest forms, that is, cloacal exstrophy [1,2]. The prenatal diagnosis of a cloacal malformation may allow better planning of pre- and perinatal care but is difficult because of the highly variable imaging features [3].

We present a rare case of cloacal anomaly, in which a cystic septate lesion in the pelvic cavity with bilateral hydroureteronephrosis and oligohydramnios with no separate visualization of the urinary bladder in a female fetus leads to suspicion of cloacal malformation which was confirmed postnatally.

CASE REPORT

A 27-year-old primigravida was referred for antenatal ultrasonography at 32 weeks of gestation. She had had three antenatal care (ANC) check-ups and two antenatal ultrasound (USG) scans at 32 and 33 weeks, respectively. No relevant medical or family history was present.

The patient had normal vitals during the ANC check-ups. Her complete blood count revealed mild anemia with a hemoglobin level of 10.2 gm/dl. The liver function test and kidney function test were within normal limits. Viral markers were negative. The patient received iron and calcium tablets during the second trimester.

On USG scanning, a large cystic lesion with midline septa measuring approximately 10×10×8 cm was noted in the fetal abdominopelvic region (Fig. 1a). The urinary tract showed bilateral hydroureteronephrosis with echogenic remnant renal parenchyma (Fig. 1b). The bladder was not visualized separately during the scan. Oligohydramnios was noted with an amniotic fluid index of 6.4. No free fluid was noted in the peritoneal cavity. Umbilical vessels were unremarkable. Repeat scanning was done a week later which showed persistence of the cystic lesion and severe oligohydramnios.

The patient delivered vaginally at 34 weeks after induction due to oligohydramnios. Postnatally, the newborn had abdominal distension with a single perineal opening between labia majora (Fig. 2a). There was rectal agenesis with no rectal opening (Fig. 2b). The APGAR score was 2 with delayed cry after resuscitation. The neonate died within the first few hours of life due to asphyxia. Consent from parents was obtained. The placental tissue was sent for aneuploidy detection which confirmed the occurrence of trisomy 21.

DISCUSSION

Cloacal malformations are rare anomalies that occur due to malformation of the urorectal septum that divides the urogenital tract from the anorectal canal [1,4,5]. The cloaca forms from the developing tail fold at 3 weeks through the confluence of the allantois and the hindgut [6,7]. The cloaca is subdivided into the

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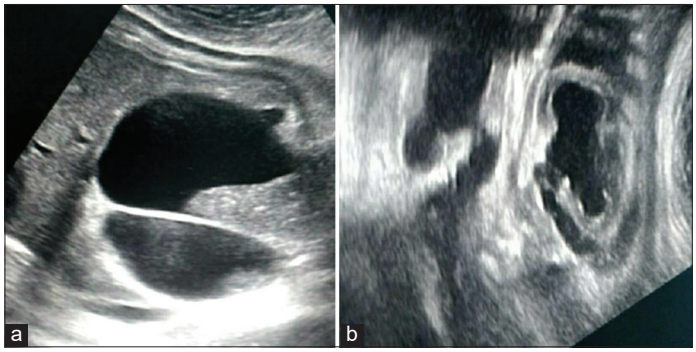

Figure 1: Ultrasound scan in coronal plane. (a) At the level of pelvis shows cystic lesion with echogenic content and midline septum. Bladder not seen separately; (b) at the level of kidneys shows bilateral hydroureteronephrosis

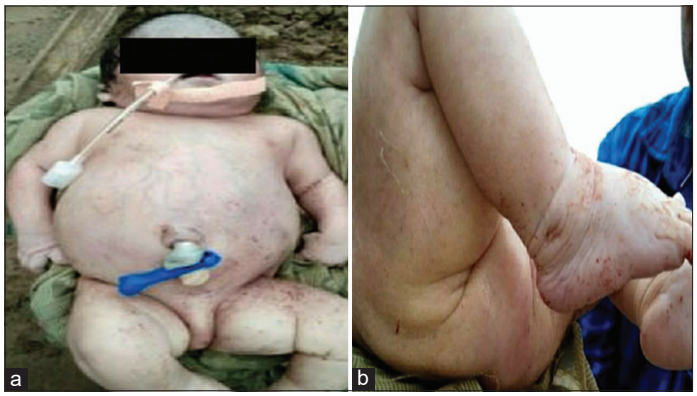

Figure 2: (a) Newborn showed abdominal distension; (b) newborn with no separate opening of vagina or anus. A single opening at site of urethral orifice was seen

urogenital sinus anteriorly and the hindgut posteriorly between the 5th and 7th weeks by the craniocaudal growth of the urorectal septum. The separate anterior urogenital sinus and posterior anorectum then open through the perineum through dissolution of the cloacal membrane once it comes into contact with the urorectal membrane.

Failure of the cloaca to subdivide leads to the persistence of the cloaca; arrest can occur at any point, leading to a wide spectrum of cloacal dysgenesis, clinically. The extent of the lesion depends on the degree of the developmental defect in the early mesoderm [6,7]. The cause of arrest is not known; however, recent evidence implicates homeobox and sonic hedgehog signaling rather than teratogenic or hereditary causes [8]. The association of Down syndrome with the artificial rupture of membrane (ARM) is well known. About 2–5% of the patients with an ARM have trisomy 21 [5,9]. However, there is no literature correlating the association of only cloacal anomaly specifically with trisomy 21. In our case, the fetus was found to have trisomy 21.

Most of the cases that have been described were diagnosed in the third trimester or immediately after birth [2,10]. The discovery of a megacyst during the first trimester may indicate a diagnosis of cloacal malformation [11]. Antenatal USG findings include transient fetal ascites, a bilobed debris-filled, abdominopelvic cystic structure, poorly or non-visualized bladder, oligohydramnios, cystic or dysplastic kidneys, and hydronephrosis. In a series of six cases of cloacal malformation that was confirmed during the postnatal period, the main prenatal USG findings were a cystic pelvic mass seen in all six cases, bilateral hydronephrosis seen in all six cases, and the lack of visible bladder was noted in three cases [3].

In our case, the cloacal anomaly presented as a cystic lesion with a median septum with no separate visualization of the bladder. It is often difficult to establish a diagnosis on USG alone. In most cases, magnetic resonance imaging (MRI) is necessary for diagnostic confirmation as communication of the cystic mass with the uterine cavity is confirmed [12,13]. Hayashi *et al*. [12] used MRI to confirm a cloacal anomaly in a fetus at 35 weeks. Warne *et al*. [3] used MRI as a diagnostic complementary method in six cases of a cloacal anomaly in the fetus between 19 and 33 weeks of gestational age. In our case, MRI was not performed because the patient was not able to afford the same.

Although USG is able to identify the large abdominal cystic mass, its origin cannot be determined in most cases. The differential diagnosis includes intestinal atresia, ovarian cysts, megabladder-microcolon-intestinal hypoperistalsis syndrome, and obstructive uropathy [2]. In the literature, cloacal anomalies have been reported to be associated with fetal ascites. Fetal urine would drain through the fallopian tube to the peritoneal cavity; this process would develop a chemical reaction that would determine the tubal obstruction, hydrocolpos, and resolution of ascites. In our case, no ascites was observed probably due to late detection by which tubal obstruction would already have occurred [2].

In our case, the presence of a septate cystic lesion in the midline abdominopelvic region in female fetus raised suspicion of a cloacal anomaly which on delivery of fetus was confirmed and the anomaly was found to be cloacal malformation with uterine didelphys with hydrometrocolpos, rectal agenesis, and bilateral hydroureteronephrosis.

CONCLUSION

The presence of a septate cystic lesion in the pelvic cavity often with a fluid-debris level with or without hydroureteronephrosis in a female fetus can guide toward making an antenatal diagnosis of cloacal anomaly. In our case, the fetus had trisomy 21 which was detected on chromosomal examination of placental tissue postnatally. The other more commonly found associations of trisomy 21 were not present in the fetus. The possible positive correlation of cloacal anomalies in trisomy 21 is a potential area of prospective research.

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