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Diagnosis of Persistent Cloaca by Ultrasonography and MRI: A Case Report

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

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Conflict of interest: None declared

Patient: Female, 38-year-old
Final Diagnosis: Persistent cloaca
Symptoms: Hydrocolpos • hydronephrosis • oligohydramnios
Medication: —
Clinical Procedure: Magnetic resonance imaging • ultrasonography
Specialty: Obstetrics and Gynecology

Objective: Congenital defects/diseases

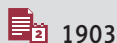
Background: Persistent cloacal malformations are rare anomalies that are anorectal malformations occurring in females. In cases of persistent cloaca, prenatal ultrasonography shows fetal ascites, cystic tumor in the abdomen, oligohydramnios, and hydronephrosis. There are various types of persistent cloaca, and symptoms vary.

Case Report: A 38-year-old pregnant woman was referred to our hospital because of suspected fetal expansion of the intestinal tract. Prenatal ultrasonography revealed a fetal growth restriction, oligohydramnios, fetal abdominal cyst, and bilateral hydronephrosis, and persistent cloaca was suspected. Also, magnetic resonance imaging (MRI) revealed a double uterus and bilateral hydronephrosis, hydrocolpos; as such, persistent cloaca was diagnosed. Cesarean section was performed at 36 weeks+3 days gestation and delivered a female infant weighing 1957 g, with Apgar scores of 9 (1 min)/9 (5 min).

Conclusions: We report a case of persistent cloaca detected in the prenatal ultrasonography and MRI examination. Prenatal diagnosis is important because it can lead to a better outcome for infants with persistent cloaca. In the image inspection in persistent cloaca, there are characteristic findings such as ascites, cystic tumor in the abdomen, difficulty in visualizing the bladder, oligohydramnios, and hydronephrosis. So, if persistent cloaca is suspected, use of ultrasonography and MRI will allow its diagnosis.

MeSH Keywords: Cloaca • Hydrocolpos • Prenatal Diagnosis

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Background

Persistent cloaca refers to anorectal malformations occurring in females. This is an extremely rare disease in which an abnormality in the formation of the urorectal septum at 5–8 weeks' gestation causes the urethra, vagina, and rectum to merge into a common tube – a cloaca – that only opens into the perineum. There are various types of persistent cloaca, and symptoms vary. Accordingly, reported fetal ultrasound findings have included fetal ascites, cystic tumor in the abdomen, difficulty in visualizing the bladder, oligohydramnios, and hydronephrosis/hydronephrosis [1,2]. We report a case involving a patient who underwent prenatal imaging and was diagnosed with persistent cloaca.

Case Report

Expansion of the intestinal tract in the fetus was revealed on ultrasound at 35 weeks' gestation, and the woman was referred to us for more detailed examination. Fetal growth restriction, oligohydramnios, fetal abdominal cyst, and bilateral hydronephrosis were observed, and persistent cloaca was suspected (Figure 1). Magnetic resonance imaging (MRI) revealed a double uterus and bilateral hydronephrosis and hydrocolpos; as such, persistent cloaca was diagnosed (Figure 2). Due to fetal distress, an emergency cesarean section was performed at 36 weeks+3 days gestation. The woman delivered a female infant weighing 1957 g, with Apgar scores of 9 (1 min)/9 (5 min). The external genitals of the baby were female. Imperforate anus and imperforate vagina were evident, and the urethral opening was unclear. The lower abdomen was remarkably distended but no other external malformations were observed. At 1 day of age, a cystostomy and a double-hole colostomy were created in the transverse colon. A contrast radiography after birth visualized the site where the 3 tracts of the bladder, hydrocolpos, and intestinal tract merged (Figure 3). In addition, a distended bladder and double uterus and hydrocolpos were found intraoperatively in the abdomen, and persistent cloaca was confirmed.

Subsequently, the infant underwent laparoscopically-assisted rectoplasty and labiaplasty. She is currently undergoing regular check-ups at our Departments of Pediatric Surgery and Urology.

Discussion

Persistent cloaca is a congenital anomaly in which the rectum, vagina, and urethra open into a common cavity due to abnormal separation of a cloaca [1,2]. A cloaca, which is a common cavity into which the hindgut and allantois open, separates into the urodeum and rectum at approximately 6 weeks'

gestation. Then, the Mullerian duct fuses with the mesonephric duct to form the uterine tract and, at the perineum, the urethra, vagina, and rectum are formed. Cloacal anomalies are a disease state in which the developmental process is impaired, and results in persistent cloaca. The incidence rate is approximately 1 per 50 000 live births [2,3]. The symptoms vary depending on the position of confluence, length of the common cavity, and other combined malformations. The higher the position of confluence, the more frequently dysuria/dyschezia occur. Approximately 40% of patients develop combined double uterus and approximately 30–50% of fetuses present hydrocolpos [2,4].

In general, prenatal diagnosis of anorectal malformations is difficult. Most cases of persistent cloaca present specific abnormalities during a fetal period and, when cysts are observed in the lower abdomen, discrimination from expansion of the intestinal tract, expansion of the renal pelvis and, in the case of a female fetus, ovarian cysts, becomes necessary. Bischoff et al. reported the frequency of abnormal prenatal ultrasound findings in patients with persistent cloaca as follows: abdominal cystic mass (41.1%), hydronephrosis (37.9%), oligohydramnios (24.2%), distended bowel (20%), ascites (15.8%), 2-vessel cord (14.7%), dilated bladder (14.7%), dilated ureter (14.7%), polyhydramnios (10.5%), echogenic bowel (8.4%), multicystic kidneys (8.4%), hydrops fetalis (7.4%), hydrocolpos (4.2%), absent kidney (3.2%), abnormal spine (3.2%), and anorectal atresia (3.2%) [5].

In general, only cystic pelvic masses in female fetuses are unlikely to be persistent cloaca. But, if there are 2 or 3 cyst structures, it is likely to be a persistent cloaca (bladder and hydrocolpos). Differential diagnosis of a female fetus with an abdominal cystic mass includes ovarian cysts, intestinal duplication cysts, cystic lymphangioma, intestinal obstruction, and small cystic colon cystic small bowel syndrome. However, by confirming the location and number of cysts, meconium, and intestinal peristalsis, it can be distinguished from persistent cloaca. Ovarian cysts are the most common cystic lesions found in the female fetus, but the cyst is lateral, not near the midline. In addition, since there is no abnormality in the anatomical structure of the kidney and the intestinal tract, it can be distinguished from persistent cloaca. Duplicate intestines are usually isolated abnormalities, but the cyst walls are thick, with multiple echogenic walls and peristalsis. An isolated midline cystic mass in a female fetus is either the bladder or the hydrocolpos. The distinction between the bladder and vagina is possible by the presence or absence of paravesical vessels in color Doppler imaging. If the cyst is in the bladder, megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is also possible. The presence or absence of meconium in the rectum is useful for discrimination from MMIHS. Meconium is present in the rectum in MMIHS, but there is no meconium

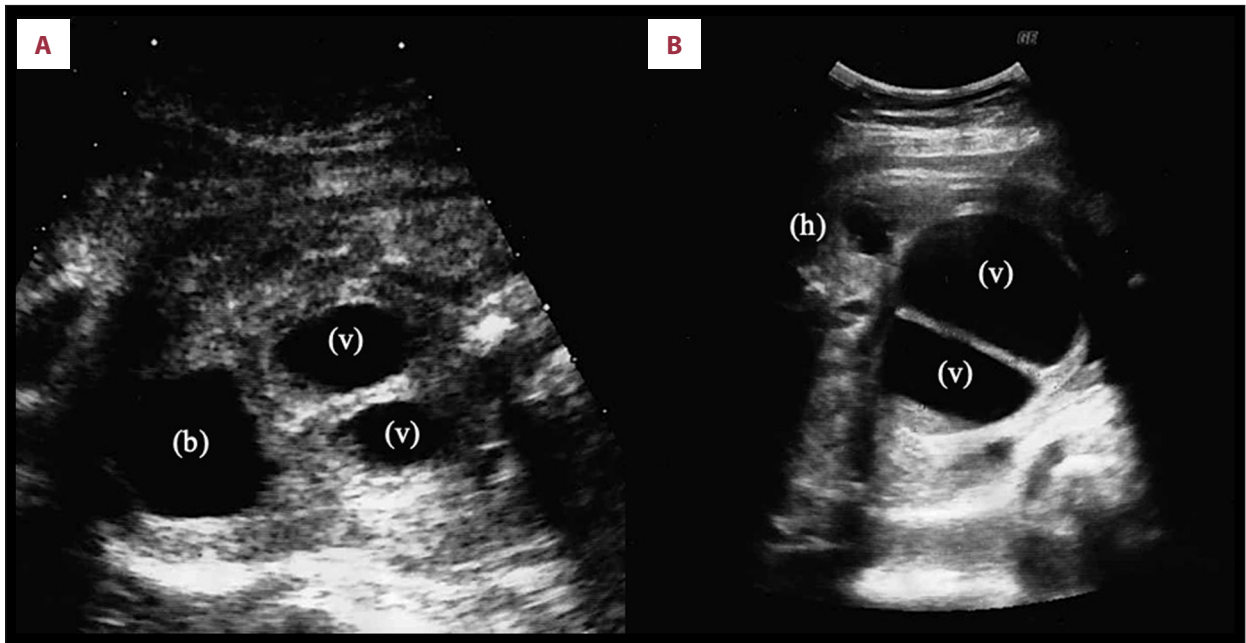


Figure 1. Ultrasound image of persistent cloaca at 35 weeks of gestation. (A) Axial image shows duplicated vagina (v), bladder (b). (B) Axial image shows distended duplicated vagina (v) and hydronephrosis (h). * Ultrasound images show multiple cysts in the abdominal cavity. The cyst is almost midline.

in the rectum in persistent cloaca. If there are 1 or 2 cysts behind the bladder, suspect hydrocolpos due to persistent cloaca. However, these may also be found in the genitourinary sinus with normal rectum or in a perforated hymen. In these diseases as well, there is meconium in the rectum in the genitourinary sinus and non-porous hymen, so it is possible to distinguish it from persistent cloaca. Based on these facts, it is considered that discrimination is possible by observing the following points carefully: 1) female fetus, 2) multiple cystic mass in the pelvis, 3) cysts near the median, and 4) no meconium in the rectum [3–5,8]. In the present case, there were characteristic ultrasonographic findings such as abdominal cystic mass and hydronephrosis, oligohydramnios, hydrocolpos. In the female fetus, there were multiple cysts near the median in the abdominal cavity, and the cloaca was easily shown by the ultrasound image (Figure 1).

Characteristic imaging findings such as fetal ascites, cystic mass, insufficient amniotic fluid, and hydronephrosis show that refluxed urine flows into the abdominal cavity through the fallopian tube, resulting in ascites and obstruction of the fallopian tube due to chronic inflammation due to urine and meconium. In 1988, Petrikovsky and colleagues reported that urine and meconium accumulate in the vagina and uterus and cause hydronephrosis [3].

In fetal ascites in persistent cloaca, urine and meconium reflux to cause meconium peritonitis. Moreover, the mixing of

urine and meconium results in calcification; therefore, intraperitoneal calcified images may be observed [6,7].

Therefore, although it is relatively easy to find persistent cloaca on ultrasound images, MRI may be helpful in diagnosis because it may show findings similar to those of other diseases. Fetal MRI has the advantage of facilitating the diagnosis of persistent cloaca by defining the anatomy of the cloaca, urogenital organs, and intestinal tracts, and has been recognized as a useful complement to ultrasound. In our case, MRI was performed, and images of bicellular cysts, hydronephrosis, and duplicated uterus were clearly shown, enabling diagnosis prenatally.

Fetal imaging assessment using ultrasound combined with MRI is important for prenatal diagnosis. In recent years, many studies have reported use of prenatal MRI. MRI has been shown to be more useful for clear visualization of lesions in the pelvis and screening for combined malformations than is ultrasound [9–12].

The optimal management of pregnancy and timing and method of delivery in cases of persistent cloaca have not been determined. When this disease is suspected, the case should be managed at a high-level facility with a department of pediatric surgery due to the possibility of other combined malformations. Because a fetus may die in the uterus due to high-grade urinary obstruction [13], and a newborn may die due to pulmonary hypoplasia and renal impairment caused by

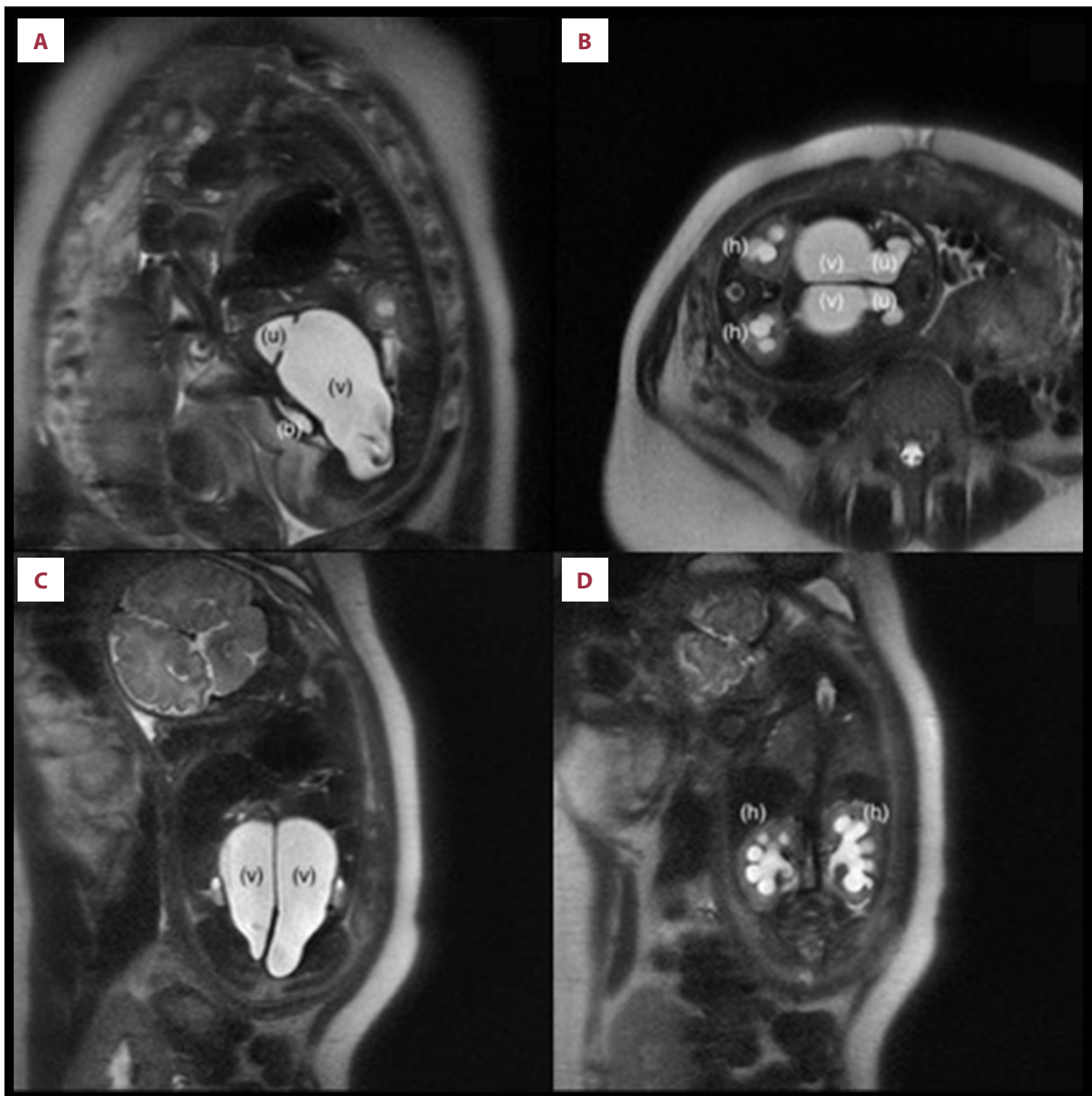


Figure 2. (A) Sagittal magnetic resonance imaging shows a markedly distended vagina (v), compressed urinary bladder (b), and dilated uterus (u). (B) Axial imaging shows distended duplicated vagina (v), duplicate uterus (u), and bilateral hydronephrosis (h). (C) Coronal imaging shows coffee-bean shaped distended duplicate vagina (v) and (D) bilateral hydronephrosis (h) caused by obstruction of both ureters.

oligohydramnios [2,3,14,15], mental health support is needed for the mother, in addition to the management of the fetus as a high-risk pregnancy [16]. Although the prognosis of persistent cloaca is not poor, persistent cloaca involves complex malformations spanning the digestive organs and the urinary and genital tracts; thus, surgical treatment and management of newborns at specialized facilities are needed. Regarding the timing and methods of delivery, delivery at term is the goal when the fetus is in good condition. However, polyhydramnios

can cause threatened premature delivery, and premature rupture of the membrane can cause preterm delivery. Although vaginal delivery is the principle delivery method, in the case of an increase in the baby's abdominal circumference, oligohydramnios, or fetal growth restriction, cesarean section is considered. In the present case, fetal growth restriction and oligohydramnios were observed. Considering the possibility of fetal distress, the patient was hospitalized and managed.

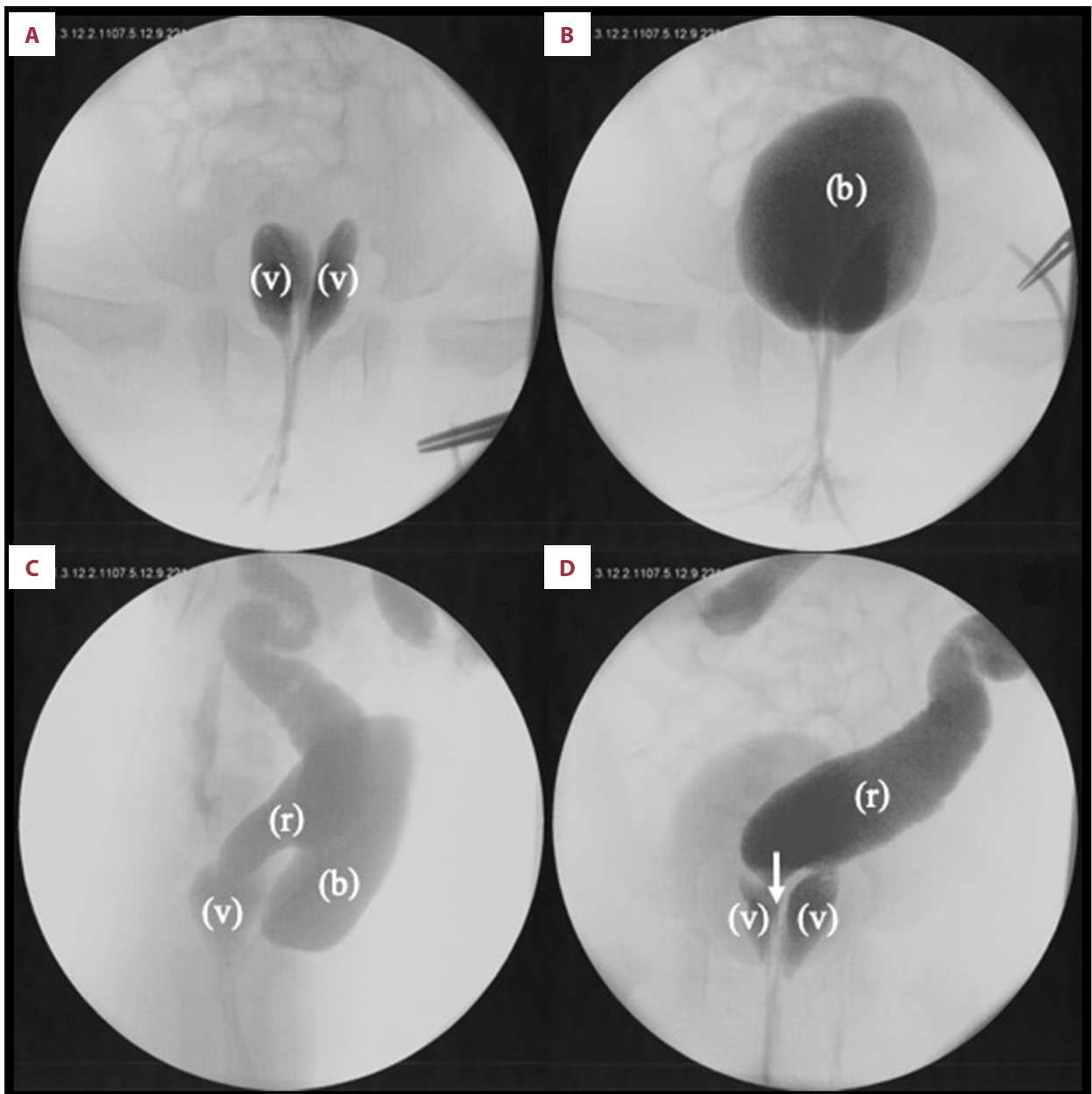


Figure 3. Contrast enema in a newborn with persistent cloaca. (A) Anteroposterior view. The image shows duplicated vagina (v). (B) The image shows bladder in front and duplicated vagina in back. (C) Sagittal view. Image shows high blind ending rectum (R). (D) Anteroposterior view. The image shows duplicated vagina (v) and rectum. The end of the intestinal tract contrasted to pass between duplicated vaginas (arrow). * Examination was performed with a Nelaton catheter placed into the bladder and vaginal cavity. Also, the rectum was imaged by inserting a Nelaton catheter from the stoma.

Cloaca is a complex malformation that spans the digestive tract, urinary tract, and genitals and requires advanced management. Therefore, based on prenatal images, information was shared in collaboration with the Neonatology Department, Pediatric Surgery, and Urology Department. The management and treatment policy should be assessed in cooperation with related departments, and explanations of the disease and mental support should be given to parents. In our case, the pregnancy

continued while carefully evaluating the fetus, but because of fetal bradycardia and fetal distress, an emergency cesarean section was performed. If fetal growth restriction and oligohydramnios are detected, it is considered a higher-risk case.

After birth, a neonatologist, pediatric surgeon, and urologist manage the child as a team [2]. Usually, drainage of hydrocolpos is performed after birth because delaying treatment with

hydrocolpos can cause urinary tract infection, pyocolpos, and vaginal perforation due to worsening urinary tract obstruction. In our case, remarkable bladder dilatation and dysuria in the common cavity were observed after birth. On the first day after birth, a colostomy and vesical fistula were performed. As a result, the ureteral compression by the expanded bladder and vagina was released, and the hydronephrosis was relieved. The bowel was dilated due to impaired defecation, and, if left untreated, abdominal distension and intestinal perforation can occur, which necessitates stoma construction after birth [5,17]. The timing of radical surgery can vary; as such, the timing depends on the type of disease, length of the cloaca, and other combined malformations. In our case, a postnatal contrast examination confirmed the urethra, rectum, and bilateral vagina opening into the common cavity (Figure 3). The child underwent vaginoplasty 1 year after birth and laparoscopically-assisted rectoplasty 2 years after birth.

Accurate diagnosis before birth and confirming detailed pathology on fetal imaging findings are useful for perinatal management and contribute to appropriate explanations to the family and improved prognosis of the child.

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Conclusions

We report a case of persistent cloaca detected by prenatal ultrasonography and MRI examination. Persistent cloaca is an anorectal malformation occurring in females. Therefore, prenatal diagnosis is important and can lead to a better outcome for infants with persistent cloaca. Ultrasonography findings in persistent cloaca are ascites, cystic tumor in the abdomen, difficulty in visualizing the bladder, oligohydramnios, and hydronephrosis. But these findings are also observed in bowel obstruction and meconium peritonitis, so differentiating these conditions from persistent cloaca is difficult. However, if the fetus is a female and there are multiple cysts near the midline in the abdominal cavity, the possibility of cloaca is high, and detailed diagnosis by MRI can be made. In our case, both MRI and ultrasonography were performed, which facilitated the diagnosis. Our experience suggests that MRI is useful for accurately diagnosing fetal persistent cloaca.

Department and Institution where work was done

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Conflicts of interest

None.