

Prenatal diagnosis of cloacal malformations

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Abstract

Introduction Prenatal diagnosis of anorectal malformations currently occurs in 0–15.9% of screened cases. In cloacas, these numbers are unknown. We speculate that some images from prenatal ultrasound studies may suggest the diagnosis of cloaca, but are not recognized because of a lack of suspicion for this diagnosis.

Methods A retrospective review of the medical records of 489 patients born with cloaca was performed; 95 of them had prenatal ultrasound reports that represent the material analyzed for this study. A literature review was performed, finding 31 publications, with 68 cloaca patients detected by prenatal images. The abnormal findings of our patients were compared with those described in the literature to determine the most common abnormal prenatal images found in patients with cloaca.

Results The 95 ultrasound reports found in our patients described 270 abnormalities, the most frequent were: abdominal/pelvic cystic/mass (39), hydronephrosis (36), oligohydramnios (23), distended bowel/bowel obstruction (19), ascites (15), 2 vessel cord (14), dilated bladder (14),

dilated ureter (14), polyhydramnios (10), echogenic bowel (8), multicystic kidney (8), “ambiguous genitalia” (7), hydrops fetalis (7), hydrocolpos (4), absent kidney (3), abnormal spine (3), and anorectal atresia (3). In spite of these findings, the radiologists who interpreted the studies only suspected a cloaca in 6 cases (6%). The literature review showed 212 abnormalities in 68 demonstrated cloaca patients. The most frequent were: abdominal/pelvic cystic/mass (46), hydronephrosis (44), ascites (21), oligohydramnios (20), distended bowel (11), multicystic dysplastic kidney (7), ambiguous genitalia (6), non-visualization of the bladder (6), two-vessel cord (5), dilated bladder (5), intraabdominal calcification (4), polyhydramnios (4), enterolithiasis (4), hydrometrocolpos (3), and dilated ureter (3).

Conclusion We conclude that it is possible to suspect the diagnosis of cloaca, prenatally, more frequently than what currently occurs, looking at the same images but with an increased index of suspicion for cystic abdominal masses and a combination of gastrointestinal and urological abnormalities.

Keywords Prenatal diagnosis · Anorectal malformation · Cloaca

Introduction

Prenatal diagnosis of an anorectal malformation occurs from 0 to 15.9% of screened cases [1–3]. In cloaca malformation specifically, these numbers are unknown.

Cloaca is one of the most complex anorectal malformation in which the urethra, vagina and rectum fuse together forming a common channel opening as a single orifice in the perineum. As much as 30% of cloacas have a

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very distended vagina, full of fluid, called a hydrocolpos, which may produce uretero-vesical obstruction, become infected (pyocolpos), and perforate. For these reasons, the hydrocolpos must be drained as early as possible after birth [4]. An accurate prenatal diagnosis of cloaca has important potential advantages for the patient, including a planned delivery at a tertiary center with experience in the management of these complex malformations and allowing for early hydrocolpos drainage in the newborn period that can avoid serious complications [4].

We speculate that some images from prenatal studies suggest the diagnosis of cloaca, but were not recognized because of a lack of suspicion for this diagnosis.

Methods

A retrospective review of the medical records of 489 patients born with cloaca and operated on by us was performed, looking for abnormalities described in their prenatal ultrasound reports.

A comprehensive literature review on the subject was performed which found 31 publications, with a total of 68 cloaca patients with prenatally detected abnormal ultrasound images [5–32]. We compared abnormal findings described in the prenatal ultrasounds of our patients with those described in the literature to find out the most common “abnormal images” in prenatal studies of patients with cloaca.

An effort was made to group abnormalities in combinations that were highly suspicious for the diagnosis of cloaca, trying to identify cases in which the diagnosis of a cloaca should have been suspected but was not.

Results

Of the 489 cloaca patients, 95 (19.4%) had ultrasound reports available, but not images. These reports described 270 abnormalities that included: abdominal/pelvic cystic/mass (39), hydronephrosis (36), oligohydramnios (23), distended bowel/bowel obstruction (19), ascites (15), two-vessel cord (14), dilated bladder (14), dilated ureter (14), polyhydramnios (10), echogenic bowel (8), multicystic kidney (8), “ambiguous genitalia” (7), hydrops fetalis (7), cloaca (6), hydrocolpos (4), absent kidney (3), abnormal spine (3), anorectal atresia (3), pulmonary hypoplasia (3), duodenal obstruction (3), ventricular septum defect (3), ureterocele (3), tetralogy of Fallot (2), anterior abdominal wall defect (2), aortic coarctation (2), calcification in the peritoneum (2), prune belly (2), and one of each: fistula between urinary and gastrointestinal tract, “ill-defined pelvic anomaly”, renal anomalies, lack of meconium in the rectum, urogenital anomaly, kidney duplication, crossed

renal ectopia, umbilical cord cyst, pericardium effusion, multiple anomalies, hepatosplenomegaly, clubfeet, shortened long bones, and cardiomegaly. Of the 39 abdominal/pelvic cystic structures, 29 were single, 7 were septated, 2 were “multiple” cystic structures, and one was triple.

There was no information on the gestational age at the time of the prenatal ultrasound reports in 64 patients. In 31 patients in whom we had information on the gestational age, the average was 27.8 weeks (range 12–37 weeks).

Literature review showed results similar to our findings with 212 abnormalities diagnosed during the prenatal period in 68 demonstrated cloaca patients. The most frequent were: abdominal/pelvic cystic/mass (46), hydronephrosis (44), ascites (21), oligohydramnios (20), distended bowel (11), multicystic dysplastic kidney (7), ambiguous genitalia (6), non-visualization of the bladder (6), two-vessel cord (5), dilated bladder (5), intraabdominal calcification (4), polyhydramnios (4), enterolithiasis (4), hydrometrocolpos (3), dilated ureter (3), and others (ventricular septal defect (3), kidney agenesis (2), pericardial effusion (2), and one of each of the following: anal atresia, anhydramnios, cloaca, esophageal atresia, deformation of the thumb, hydrops fetalis, pelvic kidney, compressed chest, sacral dimple, overriding aorta, cardiomegaly, hypoplastic bladder, septated uterus, enlarged right ventricle, tetralogy of Fallot, limb abnormality).

On adding our series’ findings to the ones in literature, the most common finding was abdominal/pelvic cystic mass present in 52% of the cases followed by hydronephrosis (49%), oligohydramnios (26%), ascites (22%), and bowel distention (18%) (Table 1).

The most common finding associated with cloacas, both in our series and in literature, was an abdominal/pelvic cystic mass. In our series, 48 reports could be interpreted as a cystic mass in the abdomen: 39 were described as a cystic mass, 5 as a dilated bladder, and 4 as hydrocolpos. Postnatally, 52 patients actually had a hydrocolpos.

Two combinations of abnormalities were found to be highly suspicious for cloaca: (1) intestinal obstruction or bowel calcifications associated with urological

Table 1 Most frequent findings of abnormalities in prenatal ultrasounds of patients with cloaca

Finding on ultrasound	Our patients (95)	Literature review (68)	Total (163)
Abdominal/pelvic cystic mass	39 (41%)	46 (67%)	85 (52%)
Hydronephrosis	36 (37.8%)	44 (64%)	80 (49%)
Oligohydramnios	23 (24%)	20 (29%)	43 (26%)
Ascites	15 (15.7%)	21 (31%)	36 (22%)
Distended bowel/bowel obstruction	19 (20%)	11 (16%)	30 (18%)

Table 2 Classification of prenatal ultrasound findings in minor, major, and highly suspicious combination for cloaca diagnosis

95 prenatal ultrasounds	36 could not have been suspected as cloacas	minor/non specific findings in prenatal US
	59 could have been suspected as cloacas (only 6 of those were suspected to be cloacas)	39 due to the presence of a major finding: abdominal/pelvic cystic mass 9 due to a combination of findings highly suspicious for cloaca in a female patient^a 5 distended bladder 4 due to the presence of hydrocolpos 2 suspected cloacas without abdominal mass^b

^a (1) Intestinal obstruction or bowel calcification associated with urological abnormalities (renal cyst, hydronephrosis, absent kidney) = 8 patients and (2) intestinal obstruction associated with abnormal spine = 1 patient

^b The other four suspected cloacas had abdominal/pelvic cystic mass

abnormalities (renal cyst, hydronephrosis, absent kidney) = 8 patients; and (2) intestinal obstruction associated with an abnormal spine = 1 patient. According to this analysis, the diagnosis of cloaca could have been suspected in 59 (62%) of our patients, instead of only 6 (6%), due to the presence of a cystic mass or one of the specific combinations of findings mentioned above (Table 2). In the 36 remaining patients without abdominal/pelvic cystic mass, the most common findings were a urologic abnormality: hydronephrosis (12), oligohydramnios (10), cystic kidneys (5), polyhydramnios (5), two-vessel cord (5), ascites (4), hydroureter (4), followed by signs of bowel obstruction or bowel calcification (5 patients).

Discussion

We are aware of the limitations of our study: we did not have access to the ultrasound images, only to the reports. We also could not have access to all of the patients with “normal” ultrasound reports. In this series, we were only exposed to the positive findings in a group of our patients. Nevertheless, we believe that this is a large series and when we compared our findings with the cases described in literature, the similarities between the results were evident and did support our findings.

We were impressed by the fact that out of 95 prenatal studies done in patients with cloacas, the diagnosis was suspected in only 6 cases. Yet, 48 of them had a midline cystic structure identified in the prenatal ultrasound (39 described as a cystic structure, 5 as a distended bladder, and 4 as hydrocolpos). Based on this series, we designed a decision-making algorithm that could help in the prenatal diagnosis of cloaca (Fig. 1). The presence of a cystic pelvic mass in a female fetus must be considered suspicious of a cloaca. The likelihood of such a patient having a cloaca

increases when two or three cystic structures can be identified (bladder and distended single vagina or septated distended vagina only, without visualization of the bladder, or bladder and distended septated vagina).

The differential diagnosis for a female fetus with an abdominal cystic mass should include: ovarian cysts, enteric duplication cysts, abdominal cystic lymphangioma, intestinal atresia, hydrocolpos due to imperforate hymen or urogenital sinus, and megacystis microcolon intestinal

Female fetus with cystic pelvic mass

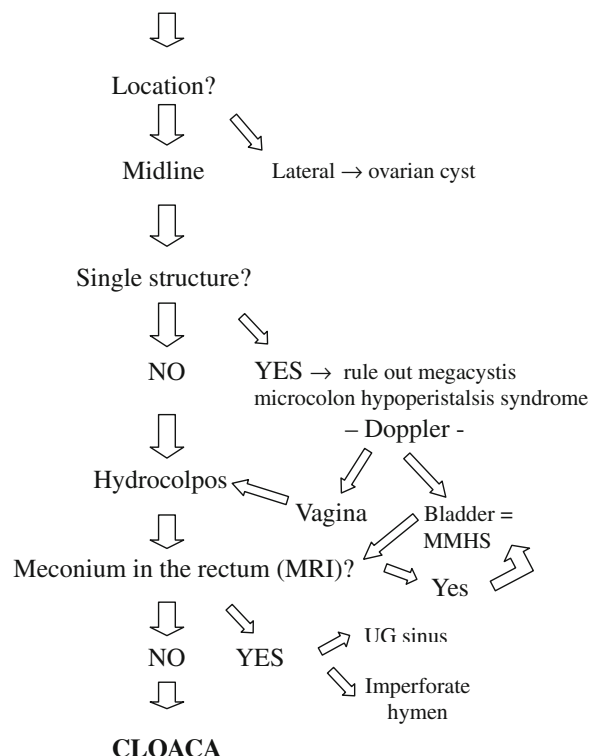


Fig. 1 Algorithm for abdominal cystic mass in female fetuses

hypoperistalsis syndrome. Other rare causes, such as urethral atresia or bladder duplication, can also be included, but are even rarer. We believe that in all cases these can be differentiated from cloaca, especially because patients with cloaca usually have associated anomalies.

Ovarian cyst is the most common cystic lesion seen in female fetuses; it is an adnexal, and lateral, rather than a midline mass. In 315 prenatal cases reported in literature [33–41], we found that there was consistently normal renal and gastrointestinal anatomy, which we believe allows the clinician to differentiate ovarian cysts from cloacas since cloacas frequently have associated anomalies.

An intestinal duplication cyst is usually also an isolated anomaly, but with a thick, multilayered echogenic wall and peristalsis in the cyst wall [42–44].

An isolated midline cystic mass in a female fetus can be either the bladder or a distended vagina (hydrocolpos). The differentiation is important, because if it is the bladder, one is obligated to rule out the possibility of megacystis microcolon intestinal hypoperistalsis syndrome (MMHS) [45]. A color Doppler might help in this differentiation, by showing the paravesical vessels [46]. If this study demonstrates that the “cystic mass” is actually the bladder, we are still obligated to look for the presence of meconium in the rectum, which will be highly suggestive of MMHS. On the other hand, the absence of meconium in the rectum will support the diagnosis of cloaca (Fig. 1).

As already mentioned, the visualization of the bladder with one or two cystic structures behind is highly suspicious of hydrocolpos due to cloaca. However, these findings can also be present in cases of urogenital sinus with normal rectum and in cases of imperforate hymen. In cases of hydrocolpos due to a urogenital sinus, there is meconium in the rectum that can be seen in magnetic resonance images. In imperforate hymen cases, there is also meconium in the rectum, and the condition seems to be less common than cloacas. A protruding membrane visible in the perineum goes with imperforate hymen.

We believe that following our algorithm in a female fetus with an abdominal cystic mass detected during the prenatal ultrasound and looking for combinations of intestinal and urological abnormalities, more cases of cloaca will be suspected during the prenatal period.

Conclusion

We conclude that it is possible to make a prenatal diagnosis of cloaca more frequently than that made currently, by looking at the same images but with an increased index of suspicion for cystic abdominal masses and a combination of gastrointestinal and urological abnormalities.

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