Seminars in Pediatric Surgery 25 (2016) 71–75

Contents lists available at ScienceDirect

Seminars in Pediatric Surgery

journal homepage: www.elsevier.com/locate/sempedsurg

Prenatal diagnosis of cloacal malformation

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ARTICLE INFO

Keywords:
Cloacal malformation
Cloaca
Anorectal malformation
Prenatal diagnosis
Ultrasound
MRI

ABSTRACT

Persistent cloaca malformation is the most severe type of anorectal and urogenital malformation. Decisions concerning the surgical treatment for this condition are taken during the first hours of life and may determine the quality of life of these patients. Thus, prenatal diagnosis becomes important for a prompt and efficient management of the fetus and newborn, and accurate counseling of the parents regarding its consequences and the future of the baby. Careful evaluation by ultrasonography, and further in-depth analysis with MRI, allow prenatal detection of characteristic findings, which can lead to diagnose or at least suspect this condition. We reviewed our experience and the literature in order to highlight the most important clues that can guide the physician in the differential diagnosis.

Persistent cloaca represents the most severe type of anorectal and urogenital malformation in which the rectum, the vagina, and the urinary tract converge into a single common channel that opens externally, at the location of the urethral meatus. Its incidence is estimated as 1 in 50,000 births.[1] The cloaca is normally present in a 4–5 week embryo until it is divided by the urorectal membrane into the urogenital sinus (anteriorly) and anorectum (posteriorly). The urogenital sinus will be further divided into the urinary system (bladder and urethra) on one side and genital (vagina) on the other side.[2] Hedgehog signaling, hereditary causes,[3] or hormonal dysregulation[4] can arrest this development at different timing, resulting in the fusion of two or more channels (urethra, vagina, and rectum). The female fetus normally presents three orifices, but in pathologic circumstances can present two perineal orifices, identified as the urogenital sinus anteriorly and the anus posteriorly, with variations in the position and integrity of these orifices. In case of cloaca malformation, instead, there is only one orifice, which can open at the site of the expected urethra or of the normal anus. The only male variant reported in literature is the cloaca dysgenesis sequence where the perineum appears smooth with no openings.[5]

Thanks to ultrasound advances, detection of fetal abnormalities in general has improved and anorectal malformations are now more commonly suspected and further investigated with more accurate imaging exams, mainly prenatal MRI.

Surgery and support care have improved in recent years. A significant number of patients have a satisfactory outcome. However, quality of life remains deeply affected in many of these patients. Thus, the advantages of antenatal diagnosis of this condition not only lie in the accurate and prompt management of the fetus and newborn, but it also improves the parental counseling to inform the parents of its consequences and the future of the baby.

Prenatal diagnosis of cloacal malformations is sustained, as most of the birth defects, by antenatal ultrasounds (US) and fetal MRI. Other imaging tools such as Doppler studies and echocardiography will add more information and will help to rule out associated anomalies.

Ultrasound findings

Persistent cloaca malformation can be present in a wide spectrum of variations, leading to different US findings (Figures 1 and 2). Although, Livingston et al.,[6] found three common signs that could raise suspicions of this condition: an intraabdominal cystic pelvic mass, urinary tract abnormalities, and dilated bowel loops,6 different authors agree that extensive ultrasound evaluation should be performed in any fetus presenting progressive enlarging cyst in the pelvis. Primary indications to guide the radiologist include undefined fetal bladder, hydronephrosis (especially if bilateral), oligohydramnios, vertebral anomalies, and ambiguous genitalia.[7]

A previous study of our group, by Bischoff et al.,[8] reports a suspected prenatal diagnosis of persistent cloaca in only 6% of a large cohort of patients (95 newborns) who actually presented this condition at birth, even though 50% presented a midline cystic structure and a total of 62% had major abnormal US prenatal findings. The authors consider the presence of a cystic pelvic mass in a female fetus as a suspicious sign for an underlying complex malformation. The specificity increases if two or three cystic structures are seen (corresponding to the bladder and the vagina, the latter frequently septated).

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http://dx.doi.org/10.1053/j.sempedsurg.2015.11.004
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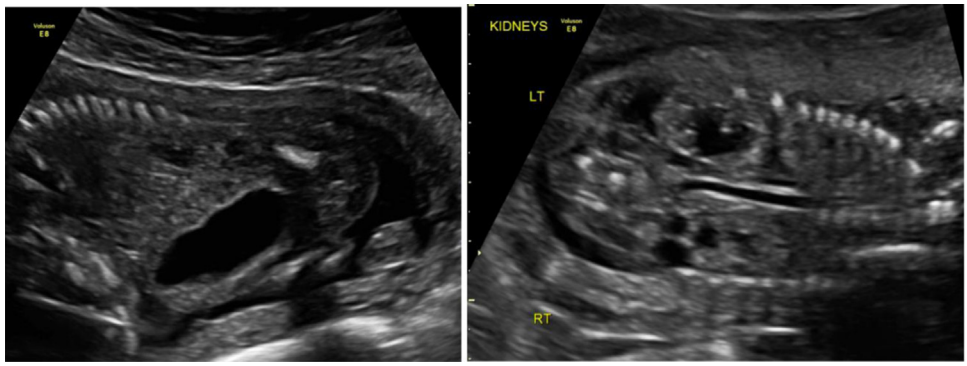

Fig. 1. Case 1. Prenatal ultrasounds at 17 weeks' gestation. (A) Large dilated cystic structure in the abdomen deforming the anterior abdominal wall. Presence of calcifications in the recto-sigmoid colon. (B) Moderate bilateral hydronephrosis. Cysts are identified in the kidneys bilaterally with perinephric fluid bilaterally that suggests calyceal rupture. Oligohydramnios.

Winkler et al.,[5] reviewed the prenatal imaging of six neonates with cloaca malformation. On ultrasound, they found a cystic pelvic mass in all their cases, with one case of septated cyst due to hydrocolpos on duplicated vaginas. They described the obstructed vagina as a median conical cyst, extended to the perineum, with a fluid–fluid level. The reduced amount of amniotic liquid depends on the mass effect of the cyst and the degree of the urinary outlet obstruction[9] that can be significant at the extreme, to produce anhydramnios and consequently severe pulmonary hypoplasia incompatible with life.[10] In case of total outlet obstruction, it has been reported that fetal ascites explained as fetal urine, drained into the peritoneal cavity, due to reflux through the fallopian tubes.[11] Peritoneal calcification can also result from meconium peritonitis if this fluid reflux contains meconium.[5]

Hydronephrosis is a common isolated finding on prenatal US, which usually resolves spontaneously after birth, but it may result from a major underlying malformation. In total, 90% of the patients with cloaca present urological abnormalities, bilateral hydronephrosis being the most common.[12] In cloaca, due to the urine outlet obstruction and the reflux to the vaginal cavity, there will be a progressive vaginal enlargement (hydrocolpos) seen in 30% of the patients, which eventually compress the adjacent structures, such as the ureterovesical junction.[1]

We can identify the following more frequent findings on the prenatal ultrasounds as suspicious signs of cloacal malformation. In order of frequency they are, abdominal cystic mass, bilateral hydronephrosis, oligohydramnios, hydrocolpos, ascites, and distended bowel.

Magnetic resonance imaging

The safety and feasibility of magnetic resonance imaging (MRI) in pregnancy has been well established and it has become a

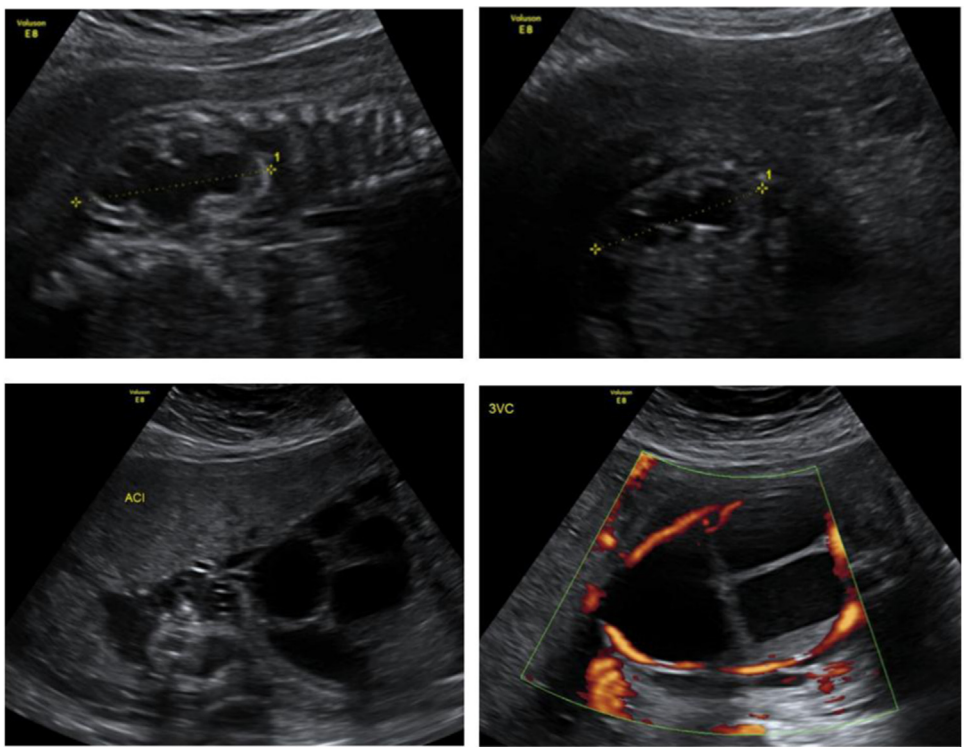

Fig. 2. Case 2. Prenatal ultrasounds at 29 weeks' gestation. Low amniotic fluid (olygohydramnios) and imaging findings most consistent with cloacal malformation. Enlarged abdomen with small lungs. Moderate to large intraabdominal fluid with calcifications and right perinephric fluid. Bilateral hydronephrosis and severely dilated bladder. Centrally septated tubular structure behind the bladder that represents hydrometrocolpos with uterine didelpys. Doppler is useful to delineate bladder position.