BMC Pregnancy and Childbirth

CASE REPORT

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Prenatal sonographic diagnosis of cloacal malformation associated with uterus didelphys and bilateral hydrometrocolpos: a case report

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Abstract

Introduction Cloacal malformation is a rare and complex group of congenital abnormalities involving anomalies of the genitourinary and gastrointestinal systems. The most common prenatal ultrasound finding is the presence of abnormal pelvic cystic masses.

Case presentation We present a case of cloacal malformation associated with uterus didelphys and bilateral hydrometrocolpos, diagnosed by prenatal ultrasound at 29 weeks of gestation. The diagnosis was confirmed by postmortem examination following induced labor.

Conclusion Abnormal pelvic cystic masses detected prenatally require close observation and monitoring. Prenatal ultrasound provides accurate and detailed diagnostic information, which is crucial to prenatal counseling and management.

Keywords Cloacal malformation, Uterus didelphys, Prenatal ultrasound, Pelvic cystic mass, Three-dimensional inversion mode

Introduction

Cloacal malformation(CM) is a rare and complex congenital condition characterized by the abnormal convergence of the rectum, vagina, and urinary tract into a single common channel, which typically opens at the site where the urethra or anus would normally be located. This condition is associated with a range of severe developmental defects, including the absence of perineal and anal openings, ambiguous vulva, as well as abnormalities in the colon, genitourinary system, lumbosacral region, and other structures. The incidence of CM is estimated to be 1 in 50,000 live births [1]. It most commonly occurs in female newborns [2],with only a small number of male cases reported in the literature [3].

Due to its rare occurrence and intricate pathological variations, the prenatal diagnosis and sonographic classification of CM remain challenging. Pena A et al. [4] classified CM into the typical cloaca and the posterior cloaca depending on the site of the opening. The opening of the former situated at the perineum, whereas the latter located near the anus. We present a case of typical CM associated with uterus didelphys and bilateral hydrometrocolpos diagnosed by prenatal ultrasound.

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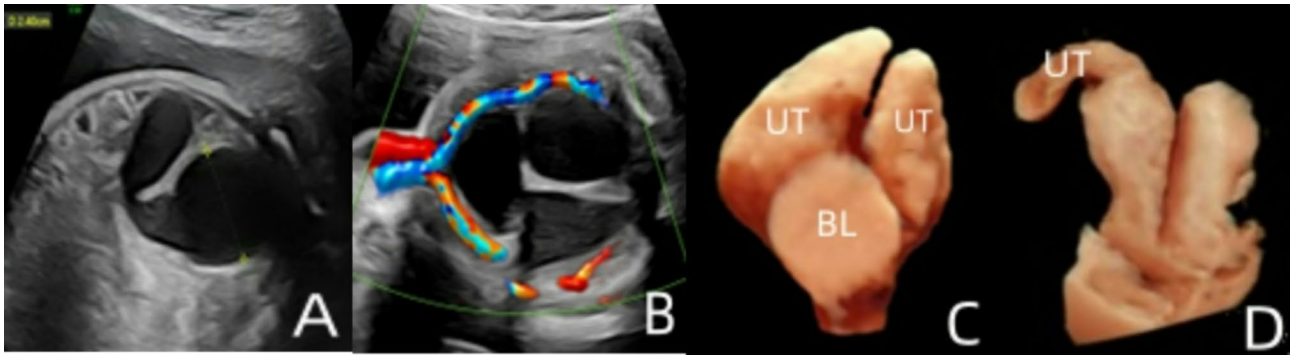

Fig. 1 Prenatal ultrasound images at 29 weeks of gestation. Two-dimensional ultrasound revealed hydrometrocolpos (**A**) and a trilobed cystic mass (**B**). Three-dimensional inversion mode imaging revealed bilateral hydrometrocolpos (**C**) and one of the uterine horns in the uterus didelphys (**D**). UT, uterus; BL, bladder

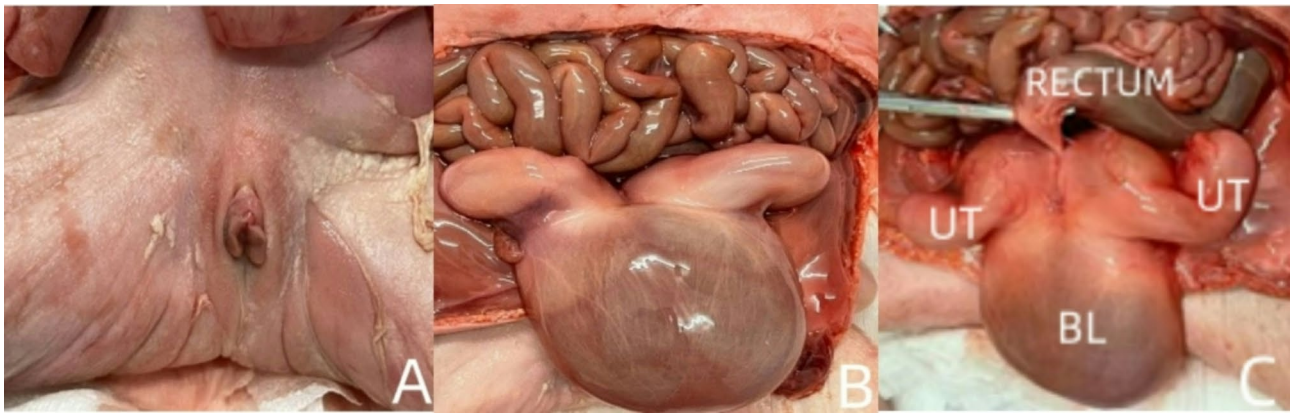

Fig. 2 Postmortem examination images. A stillborn female infant was delivered without an anus, with only one opening was observed in the perineal vestibule (**A**). Uterus didelphys was noted posterior to the bladder (**B**). Bilateral hydrometrocolpos and terminal rectal atresia were observed (**C**)

Case presentation

A 36-year-old woman, gravida 2para 0, with an unremarkable medical history and no significant family genetic background underwent a routine prenatal ultrasound at 29 weeks of gestation. The ultrasound revealed a single fetus in a breech presentation. The fetal biometry parameters were consistent with the gestational age, and the amniotic fluid index measured 15 cm. The fetal left kidney appeared normal. The right kidney's collecting system was separated by approximately 1.0 cm, and the inner diameter of the right ureter measured about 0.2 cm. The bladder size was approximately 1.5 × 1.2 cm. A bilobed cystic mass was observed posterior to the bladder, measuring approximately 5.4 × 2.3 cm and 6.4 × 2.2 cm. The proximal portion demonstrated bilateral horn-like configurations (Fig. 1A), while the distal aspect appeared contiguous with the bladder wall. Numerous hypoechoic foci were observed within the cystic mass. These components collectively formed a trilobed cystic complex (Fig. 1B). On dynamic imaging, the bilobed cystic mass exhibited mild peristaltic and antiperistaltic movements without significant morphological alterations. The echogenicity of this region was distinctly different from adjacent bowel loops and the urinary bladder. No appreciable dilation was observed in the intra-abdominal bowel loops. The fetal anal target sign (sphincter complex) and anorectal canal remained unidentified. External genitalia demonstrated typical female morphology. Three-dimensional inversion mode imaging revealed bilateral hydrometrocolpos (Fig. 1C) and one of the uterine horns in the uterus didelphys (Fig. 1D). Fetal cloacal malformation associated with uterus didelphys and bilateral hydrometrocolpos was diagnosed by prenatal ultrasound. Three days later, the pregnant woman did not perceive any fetal movement, and the follow-up ultrasound indicated stillbirth. A stillborn female infant without an anus was delivered vaginally at 30 weeks of gestation following labor induction. Only one opening was observed in the perineal vestibule (Fig. 2A). The prenatal ultrasound diagnosis was confirmed by postmortem examination. Postmortem examination revealed a uterus didelphys positioned posterior to the bladder, concurrent bilateral hydrometrocolpos and terminal rectal atresia (Fig. 2B, C).

Discussion

During the third week of embryonic development, the hindgut joins with the allantois to form the cloaca. Between the fourth and seventh weeks, the urorectal septum and cloacal membrane collaborate to partition the cloaca into the ventral urogenital sinus and the dorsal anorectum. The former gradually differentiates into the genitalia, bladder and urethra, while the latter develops into the distal rectum, anal canal and anus [5, 6]. Currently, the pathogenesis of CM remains unclear. It is hypothesized that failure of the urorectal septum to properly separate the cloaca, or its inability to fuse with the cloaca membrane, may contribute to simultaneous disturbances in the differentiation of internal and external genital [7]. Additionally, the pathogenesis may be associated with genetic mutations that lead to mesodermal defects, as well as exposure to teratogens, including chemicals, viral infections, and adverse environmental factors [8].The genes and molecular pathways governing the differentiation and development of the hindgut and lower urinary tract have been extensively explored, with Sonic hedgehog (Shh) signaling identified as a critical regulator [9].

CM manifests as a complex spectrum of rare and severe multisystem malformations that lack distinct sonographic features, making the prenatal diagnosis of urogenital anomalies highly challenging. Livingston et al. [10] proposed a diagnostic triad for CM comprising an intra-abdominal cystic pelvic mass, urinary tract abnormalities, and enteric dilatation. While ultrasonographic imaging demonstrate high sensitivity in detecting cystic pelvic lesions, definitive tissue origin determination remains elusive [11].

In our case, the fetal pelvic trilobed cyst raised clinical suspicion. The distinctive morphology, mild peristalsis, and relative rarity of cyst frequently result in their misdiagnosis as intestinal dilation. When detecting cystic pelvic masses, a comprehensive diagnostic approach should include: systematic evaluation of morphological features, comparative analysis of intracystic echo patterns against bladder and intestinal lumina, and dynamic assessment of spatial relationships with adjacent anatomical structures, including the bladder, urethra, rectum, anal canal, uterus, and vagina. Concurrent examination of external genitalia and anal target sign provides critical diagnostic clues. Three-dimensional inversion mode imaging further enhances diagnostic precision by three-dimensionally visualizing morphology and spatial configurations, serving as a valuable adjunct for clinical decision-making.

Conclusion

The presence of an abnormal pelvic cystic mass serves as a suspicious indicator of complex pelvic malformations, necessitating cautious observation and constant monitoring. CM encompasses a range of severe congenital defects with a poor prognosis, and affected infants often need multiple genitourinary and bowel reconstruction surgeries. Consequently, prenatal ultrasound provides critical diagnostic insights, playing a vital role in guiding prenatal counseling. In addition, the application of three-dimensional inversion mode imaging enhances clinical information, serving as a valuable supplementary tool.

Abbreviations

CM	Cloacal Malformation
UT	Uterus
BL	Bladder
Shh	Sonic Hedgehog

Acknowledgements

We are grateful for the participation of the patient from whom we drew the clinical information in this report and who agreed to the publication of this information.

Author contributions

XX contributed to data collection, manuscript writing and revision.RT and XL prepared Figs. 1 and 2. LL contributed to data collection.HY contributed to revising the manuscript.All authors reviewed the manuscript.

Funding

This study was supported by Clinical Medical Technology Demonstration Base for Genetic Research of Fetal Congenital Heart Disease in Hunan Province (2021SK4036) and Hunan Province Children's Safe Medication Clinical Medical Technology Demonstration Base(2023SK4083). The funding body do not have any role in the design of the study, the collection, analysis, and interpretation of the data, and in writing the manuscript.

Data availability

All data generated or analyzed during this study are included in this article. Further enquiries can be directed to the corresponding author.

Declarations

Ethics approval and consent to participate

This study was approved by the Human Research Ethics Committee of Changsha Maternal and Child Health Hospital.
All the authors approved the manuscript.

Consent for publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Competing interests

The authors declare no competing interests.

Clinical trial

Not applicable.

Received: 8 May 2024 / Accepted: 26 March 2025
Published online: 04 April 2025

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