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Imperforate hymen with renal complications? A case of 2-day-old patient with hydrometrocolpos

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Abstract

Hydrometrocolpos as is a rare condition in female newborns which leads to the accumulation of fluid above the obstruction. Its most common causes are imperforated hymen or vaginal abnormalities.

A premature baby born at 33+2 weeks was found to have generalized body oedema. Foetal MRI revealed hydrometrocolpos with imperforated hymen and ureteral compression, which led to dilatation of the left renal pelvis. USG (ultrasonography) revealed that the patient also had free fluid in the abdominal cavity. A significantly enlarged vagina and uterus was seen extending to the bottom of the stomach. The patient was qualified for surgery, which involved incising the hymen and evacuating the watery-mucus contents of the vagina. Early diagnosis of hydrometrocolpos using ultrasound or MRI is crucial to detect other serious abnormalities.

Key words

hydrometrocolpos, urogenital disease, imperforate hymen, vaginal obstruction

INTRODUCTION

Hydrometrocolpos (HMC) is the accumulation of fluid in the uterine and vaginal cavity, which leads to their extension, with nearly 100% diagnostic certainty achievable from the prenatal period to pre-pubertal age [1, 2, 3]. In pubertal patients, differential diagnoses include both hydrometrocolpos and haematometrocolpos. This rare congenital condition has an incidence of about 0.006%, with occurrence rates in full-term newborns ranging from 0.0014% – 0.1% [1]. A recent review (2018–2023) identified 69 cases, mostly diagnosed prenatally or shortly after birth [2].

HMC typically results from congenital factors, with an imperforate hymen being the most common cause. Imperforate hymen occurs due to incomplete resorption of the hymenal membrane during embryonic development. Other causes include transverse vaginal septum, distal vaginal atresia, or associations with anorectal anomalies. There are two types of hydrometrocolpos, categorized by the type of fluid that accumulates: the urinary type, connected with cloacal anomaly or persistent urogenital sinus, and the second type, which is associated with increased maternal estrogenic stimulation, leading to the production and secretion by the uterine and cervical glands in the foetus and newborn [4, 5, 6].

Complicated cases in newborns require urgent management to avoid severe complications, e.g. sepsis or renal failure [4]. Prenatal ultrasound is the key to diagnosis, revealing a cystic structure behind the bladder and anterior to the rectum. Magnetic resonance imaging (MRI) may be helpful to confirm the diagnosis [3, 5]. The size and location of the cyst depend on its cause and severity. Although some cases are diagnosed as early as 25 weeks of pregnancy, most prenatal diagnoses occur in the later weeks of the second or third trimester. Early detection is critical for planning effective postnatal interventions [2].

The case is presented in which prenatal ultrasound and prenatal magnetic resonance imaging was critical in making the diagnosis of hydrometrocolpos.

CASE REPORT

A mother at 31 weeks of pregnancy reported to the hospital due to chronic and persistent cough following an influenza type A infection two weeks previously. During hospitalization, a routine foetal ultrasound was performed which revealed the presence of a fluid-filled lesion in the pelvic area measuring 20 × 30 mm, free fluid measuring up to 23 mm in the Morrison pouch, and dilation up to 8 mm of the calyceal-pelvic system of the left kidney. Suspecting an ovarian cyst or hydrosalpinx, the attending physician recommended a foetal MRI.

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One week later, at 32 weeks of pregnancy, MRI was performed and revealed hydrometrocolpos. The uterus and vagina were significantly distended and filled with homogeneous fluid. The fetal vagina measured 76 × 38 × 38 mm and the uterus – 45 x 46 x 38 mm. The calyceal-pelvic systems of both kidneys were dilated along with the ureters. Diameter of the left ureter was up to 6 mm and of the right ureter up to 4 mm.

At 33 weeks of pregnancy, due to premature rupture of membranes and the onset of premature labour, the decision was made to perform a Caesarean section. The baby girl was born with severe generalized oedema, labial swelling, signs of hypertrophy (body weight at the 97th percentile), and ascites. The Apgar scores of the newborn were 6/6/7/8. After birth, the newborn was placed under a radiant heater, dried, given five resuscitation breaths, and then ventilated. After extraction, she passed urine and meconium. Due to symptoms of respiratory distress syndrome, duo positive airway pressure (DUOPAP) was applied at the tenth minute of life, and the decision made to transfer the newborn to the Paediatric Intensive Care Unit (PICU) at the University Children's Hospital for further diagnosis and treatment. On admission to the PICU the girl was intubated and ventilated, her skin was reddened with cyanosis of the extremities, and generalized body oedema. Vital signs were measured: blood pressure: 47/23 mmHg, capillary refill time: 4–5 sec. Chest X-ray revealed RDS (Respiratory Distress Syndrome). Exogenous surfactant (Curosurf®) was administered. intratracheally. Cardiac output was supported by dobutamine and dopamine infusion, and blood pressure normalization was achieved at 69/39 mmHg.

During the initial gynecological consultation, an imperforate hymen was suspected; however, the girl was not qualified for urgent surgical intervention due to the absence of fluid in the uterus and vagina on the first postnatal ultrasound performed. Surgical procedure was deferred until other causes of ascites could be determined. The next day, another ultrasound examination revealed the presence of free fluid in the abdominal cavity which was visualized in the hepatorenal and splenorenal recesses. In addition, bilateral dilation of the calyceal-pelvic systems, enlarged vagina with a diameter of 16 mm, and a uterus with a fundus extending up to the stomach and filled with dense contents, were documented (Fig. 1).

The patient was qualified for urgent surgery, during which the imperforate hymen was incised, allowing for the evacuation of the accumulated watery-mucous content

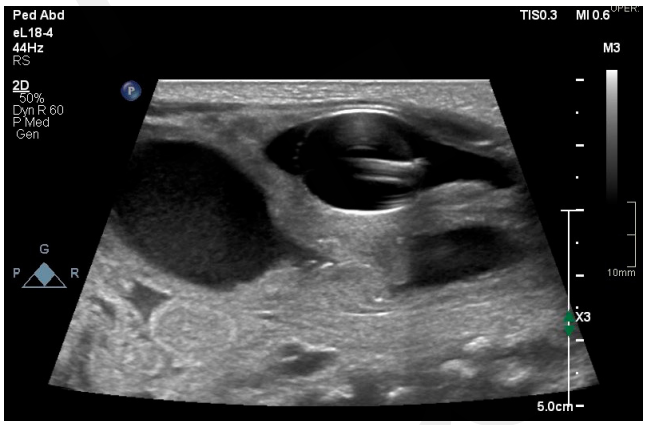

Figure 1. Ultrasound photo showing dilated vagina and uterus filled with dense content

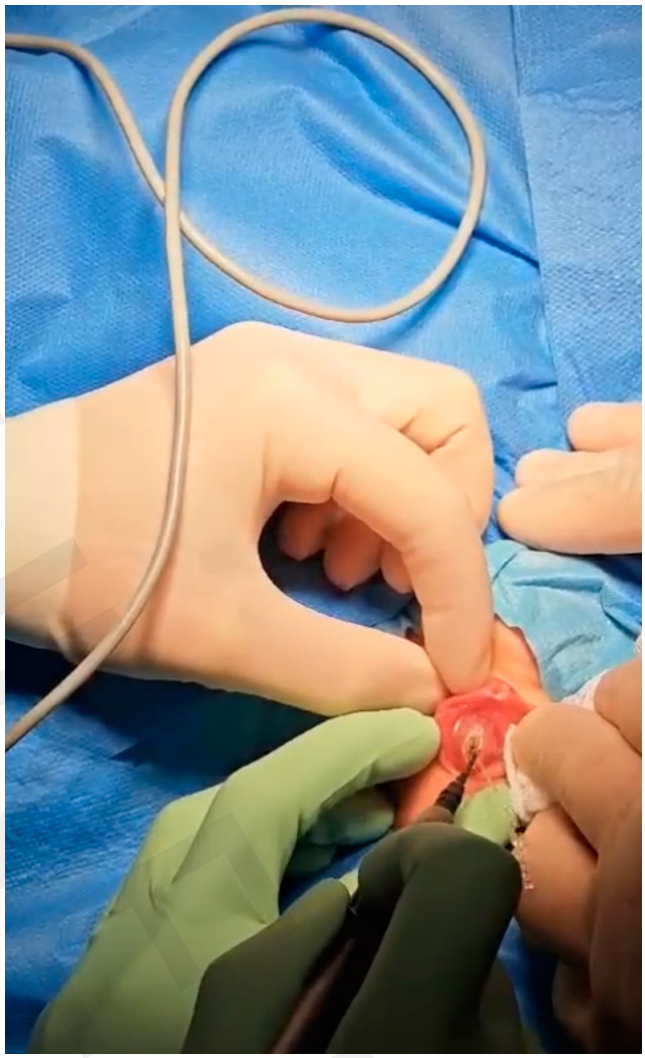

Figure 2. Moment of incision of the imperforate hymen and evacuation of mucous-watery content from the vagina

(Fig. 2). Subsequently, the hymen was reconstructed to maintain the patency of the vaginal canal. Additionally, a cystoscopy was performed – the bladder was slightly deformed by the previously distended uterus and vagina. However, the patency of the ureteral orifices was normal. After surgery, the girl was in good general condition and transferred to the Neonatal Pathology Department, where she was placed in an incubator and monitored. Treatment continued from the Intensive Care Unit.

Post-operative ultrasound revealed that all previously dilated structures had returned to normal dimensions, and no fluid was detected in the abdominal cavity. A follow-up gynecological consultation two weeks after the procedure confirmed the normal anatomy of the vaginal vestibule, normal labia, a non-enlarged clitoris, and maintained patency of the hymen.

The patient was discharged from hospital six weeks after birth. During follow-up consultation two months later, patency of the hymen was still present and an ultrasound of the reproductive organs was normal for the girl's age. Genetic testing performed with the CytoScan 750K array revealed normal balanced female karyotype (arr (X,1–22)x2).

DISCUSSION

Congenital imperforate hymen, the most common obstructive anomaly of the female reproductive tract, is rarely diagnosed in neonates. It is typically identified in pubertal girls presenting with amenorrhea, pelvic pain, and abdominopelvic mass [4, 5]. In neonates, hydrometrocolpos can cause severe symptoms due to compression of vital organs. Increased abdominal pressure from the pelvic mass may lead to respiratory failure requiring intensive care, while urethral obstruction can result in kidney dysfunction [7]. Gastrointestinal complications, such as peritonitis or acute intestinal obstruction, may also occur. Additionally, venostasis in the lower limbs, causing oedema, has been reported [2]. Early diagnosis and intervention are critical to prevent these potentially life-threatening complications. Some of these severe symptoms occurred in the patient presented in the current case report. In the presented case, the girl was born with signs of respiratory distress and needed intensive care in the PICU, including synchronized intermittent mandatory ventilation (SIMV) and exogenous surfactant.

In the presented case, the etiology of the patient's RRS (Rapid Response System) is not precisely known. It could be related to premature birth age, congenital pneumonia or diagnosed hydrometrocolpos. She presented with generalized oedema, ileus, and water-electrolyte and acid-base imbalances, accompanied by haemodynamic instability that needed immediate management with inotropic drugs.

Influenza infection during pregnancy can be associated with complications in foetal development [8]. The non-systematic data search performed using three large databases: Pubmed, Scopus and Embase did not return any reports linking influenza infection and congenital reproductive tract anomalies. The majority of papers focused on the association of maternal influenza with congenital heart defects and neural tube defects [9]. On the other hand, an increased risk of preterm delivery was connected to both the influenza infection during pregnancy and hydrometrocolpos. A recent review on hydrometrocolpos highlighted a series of three patients with that anomaly, who were born prematurely at 29, 34, and 36 weeks of gestation [10]. In the presented case, the mother was hospitalized in her 31st week of pregnancy for a prolonged cough, during which a routine ultrasound revealed a fluid-filled lesion. Early imaging diagnostics were crucial in preventing complications, such as sepsis, intestinal obstruction, or renal failure. The case was complex due to the mother's infection, premature labour caused by membrane rupture, and the newborn's poor condition with generalized oedema. Despite noting an imperforate hymen on the first postnatal day, surgery was initially deferred as the uterus was not fluid-filled, and other causes for the neonate's distress were considered. However, on the second postnatal day, ultrasound monitoring revealed fluid accumulation in the uterus and vagina, leading to urgent hymenal surgery. Postoperative recovery confirmed that hydrometrocolpos was the primary cause of the newborn's distress, rather than prematurity or maternal infection.

Hydrometrocolpos in neonates, although rare, is often associated with such congenital anomalies as polydactyly, which may indicate genetic syndromes such as McKusick-Kaufman syndrome (MKKS), Bardet-Biedl syndrome, or Ellis-Van Creveld syndrome [5, 6]. In a review by Grant et al., seven of 69 cases (10%) were linked to MKKS [2]. While the current patient lacked anomalies indicative of MKKS, genetic testing was performed. The results of genetic testing did not reveal any abnormalities that could suggest a genetic etiology of this pathology in the patient. Differential diagnosis should consider obstructed hemivagina and ipsilateral renal anomaly (OHVIRA), found in 20 of 69 reviewed cases [2]. Importantly, HMC should be evaluated when diagnosing cystic abdominal lesions, irrespective of other symptoms. Early recognition is crucial for appropriate management and preventing complications.

CONCLUSIONS

This case highlights an important causal association between imperforate hymen – commonly regarded as a benign condition – and the occurrence of severe life-threatening complications in premature newborns. It also underlines the necessity for meticulous inspection of the genital area which is fundamental for comprehensive care of patients with hydrometrocolpos. A key element in diagnostics is performing a prenatal ultrasound, as well as considering foetal MRI. It is also prudent to consider that this condition may be a sign of genetic syndromes, such as McKusick-Kaufman, MRKH, OHVIRA or Currarino syndrome. Early diagnosis of both hydrometrocolpos and other associated anomalies is crucial for initiation of appropriate treatment and prevention of serious complications.

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