

Isolated Foetal Ascites Associated with a Urachal Cyst and Posterior Urethral Valves: A Case Report and Literature Review

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Keywords

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Abstract

Background

Prenatal detection of isolated ascites in combination with an abdominal cyst poses a diagnostic challenge. The diagnosis of urachal remnants may be particularly difficult, as imaging findings can mimic other urinary tract anomalies. Furthermore, their rarity has resulted in a lack of clear evidence-based management guidelines.

Methods

A literature review was conducted to understand this clinical presentation. The Medline database (PubMed) was searched using the MeSH terms "Ascites/congenital," "Urachal Cyst," and "Urethral Obstruction/congenital." A total of 979 publications were screened.

Results

Urachal cysts are rare embryological remnants that are often asymptomatic and may regress spontaneously, supporting conservative management in selected paediatric cases. In contrast, posterior urethral valves require prompt diagnosis using voiding cystourethrography, followed by valve ablation. Long-term follow-up of these patients is essential, as up to 20% of affected patients may progress to end-stage renal disease.

Discussion

Early recognition of this rare combination of findings may facilitate timely diagnosis of posterior urethral valves and guide appropriate post-natal management. In this case, the urachal cyst may have acted as a pressure-relieving "pop-off" mechanism. This might explain the absence of hydronephrosis and could have potentially protected the renal function. Clinical follow-up will be necessary to determine whether this presumed pressure-relief mechanism translates into a more favourable long-term renal prognosis.

Introduction

This report describes the unique presentation of a neonate with antenatally detected isolated ascites and bladder wall thickening in combination with an abdominal cyst. Prenatal imaging suggested a urachal cyst, given its position between the bladder dome, and the anterior abdominal wall at the level of the umbilical insertion. However, the diagnosis of urachal remnants remains challenging, as imaging findings may resemble other urinary tract anomalies. The associated ascites and bladder wall thickening raised suspicion of uroperitoneum secondary to a suspected lower urinary tract obstruction. During further diagnostic evaluation, this neonate was eventually diagnosed with posterior urethral valves (PUV), a finding that was essential for therapeutic decision making and clinical follow-up. The coexistence of these conditions suggests a potential pathophysiological relationship, which will be further explained in this report. A narrative review of the literature was performed to further

analyse this clinical presentation, with particular emphasis on the prenatal presentation of intra-abdominal cysts, the differential diagnosis of isolated ascites and the current evidence regarding the therapeutic management of posterior urethral valves and urachal cysts. The aim of this case representation is to guide clinicians in the diagnostic process, as a timely diagnosis is crucial in these patients to support optimal medical management.

Case presentation

The mother of this patient was referred to a tertiary hospital at 33 weeks of pregnancy due to the presence of isolated foetal ascites. Until then, the pregnancy had been uncomplicated. The family history was negative and the parents were non-consanguineous. Specialist ultrasound showed clear ascites and a cystic echolucent structure between the bladder and umbilical cord insertion (Figure 1). The bladder wall appeared thickened, without apparent keyhole sign,

TABLE 1: Comparison of biochemical parameters in blood, urine, and ascitic fluid on day 1.

	Blood	Urine	Ascites	Unit
Glucose	68	Negative	84	mg/dL
Albumin	38	/	12,4	g/L
Creatinine	0,91	20,2	0,90	mg/dL
Leukocytes		105	64	/microL
Erythrocytes		12	<1000	/microL

and there was a normal appearance of both kidneys without hydronephrosis. The anal canal and rectum were visible and appeared normal. Cardiac function was normal, with no evidence of hydrops fetalis or foetal anaemia. There were no features suggestive of a lymphatic disorder. The differential diagnosis included urinary ascites (secondary to bladder rupture or rupture of a urachal cyst), and, less likely, intestinal perforation secondary to bowel obstruction. A primary caesarean section was performed at 36 weeks 4 days of gestation because of breech presentation and anticipated respiratory compromise resulting from increased intra-abdominal pressure, as reflected by diaphragmatic elevation. Antenatal corticosteroids were administered to minimize the risk of transient tachypnoea of the neonate. The neonate demonstrated a good adaptation, showed no dysmorphic features and had a birthweight of 3000 grams (percentile 61). Spontaneous micturition was observed immediately after birth, although the quality of the urinary flow was not documented. The neonate was intubated because of restricted lung expansion due to the abdominal distention. An indwelling catheter was placed after birth by the paediatric urologist. Initial urine output showed transient polyuria during the first hours (6,3 mL/kg/hour), which subsequently normalized (approximately 3 mL/kg/hour). Due to abdominal distension causing respiratory compromise, paracentesis was performed, draining 350–400 mL of ascitic fluid. Following paracentesis, the infant was successfully extubated on day 2. Analysis of the ascitic fluid demonstrated a creatinine concentration comparable to serum (Table 1). Postnatal ultrasonography demonstrated a thickened bladder wall and an anechoic cyst anterior to the bladder consistent with a urachal cyst, without other abnormalities of the urinary tract (Figure 2). Initial voiding cystography on day 1 excluded urinary leakage or bladder perforation. The ascites drain was clamped on day 2, after stabilization of the abdominal circumference, and removed on day 7, with no recurrence of ascites during follow-up. Enteral feeding was initiated on day 4, with spontaneous meconium passage, and full enteral feeding was achieved by day 8. After starting enteral feeding, chylomicrons were not detected. MRI of the abdomen on day 5 confirmed the urachal cyst and bladder wall thickening, without additional abnormalities (Figures 3a and 3b). Biochemical analyses showed normal

values for transaminases, coagulation, albumin and ferritin (differential diagnosis of gestational alloimmune liver disease). Metabolic testing showed normal results. Additionally, molecular karyotyping and a targeted metabolic gene panel were performed, both yielding normal results. Repeat voiding cystourethrography on day 7 demonstrated dilatation of the posterior urethra proximal to the level of obstruction suggestive of PUV, with active vesicoureteral reflux grade 1. Subsequently, a cystoscopy on day 10 confirmed the presence of PUV and a urachal opening with a cystic remnant. Valve ablation was postponed because of a narrow urethral meatus. The neonate was discharged on day 12 with an indwelling urinary catheter and antibiotic prophylaxis. At the age of 5 weeks, endoscopic resection of the PUV was successfully performed, after which the catheter could be removed. The patient is regularly seen by the paediatric nephrologist and urologist. As of the latest follow-up, there have been no episodes of urinary tract infection.. Follow-up ultrasonography demonstrated persistent bladder wall thickening without hydronephrosis. Biochemical parameters remained within the normal range. During a second cystoscopy at four months of age, no residual urethral valves were observed; only a small cyst was seen in the median cranial portion of the bladder.

Discussion

Antenatal diagnostic work-up and follow-up

In this case the antenatally diagnostic process was based on the visualization of an abdominal cyst in combination with isolated ascites. Most abdominal cystic lesions are detected during the second or third trimester of pregnancy (1). To further optimize the diagnostic workup of a foetal abdominal cyst, several important considerations should be taken into account. An important remark is that a pelvic cyst seen early during gestation, could be a sign of a dilated distal rectum in foetuses with an anal atresia. Therefore, careful evaluation of the perianal muscular complex remains essential to exclude the possibility of an anal atresia (2). The differential diagnosis of foetal abdominal cysts is summarized in Table 2 (1,3,4). In this case a urachus cyst was already suspected antenatally due to the anatomical relationship between the cyst, the anterior abdominal wall at the level of

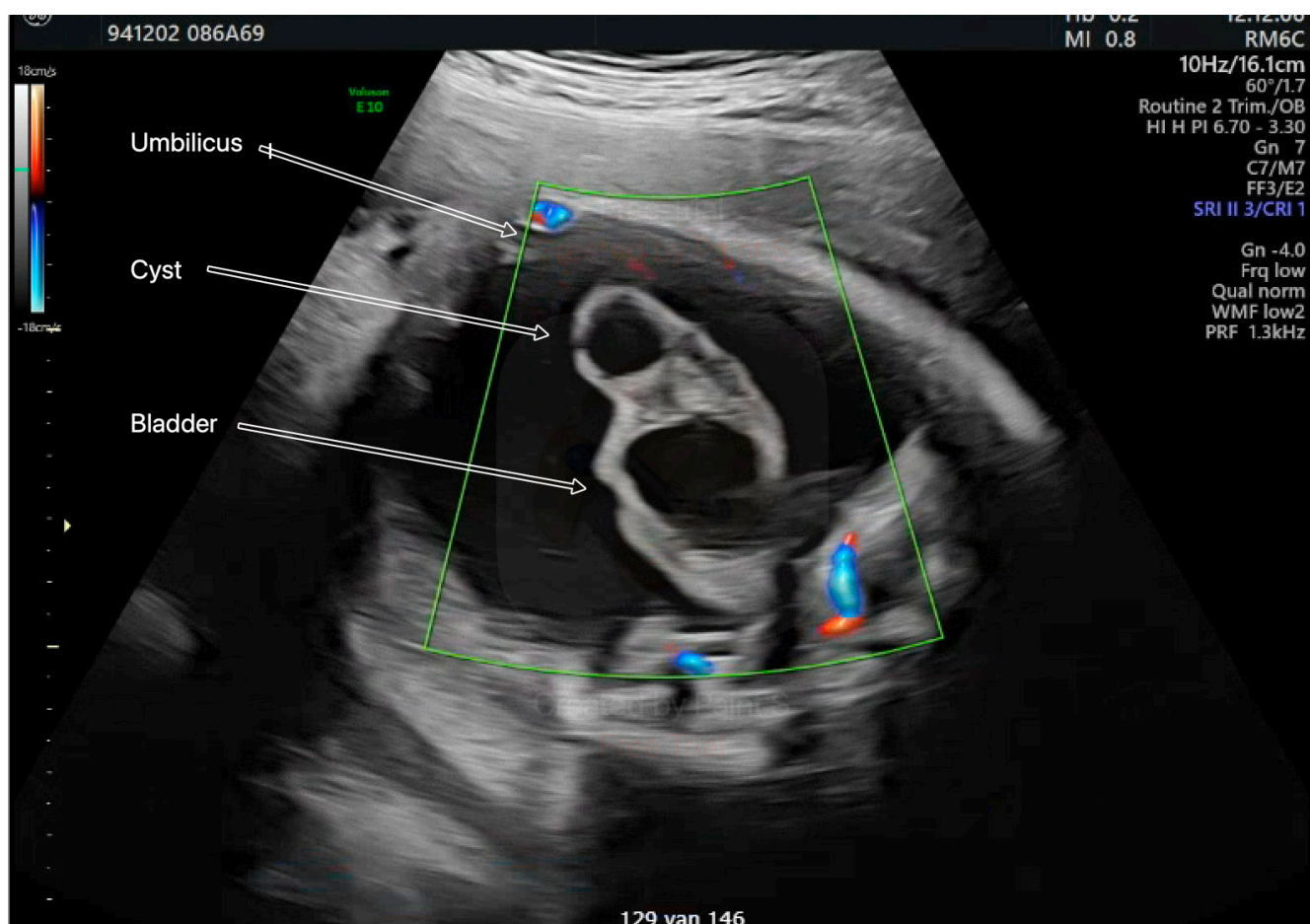


FIGURE 1: Antenatal ultrasound with abdominal cyst (between the bladder wall and the umbilical cord insertion), thickened bladder wall and clear ascites.

the umbilical insertion, and the bladder. The urachus represents an embryological connection between the bladder and the allantois, which normally undergoes obliteration during foetal development or in the early neonatal period (5). Failure of this involution results in the persistence of a urachal remnant, which may present as one of the following anomalies: a patent urachus with a persistent lumen between the bladder and umbilicus; a vesico-urachal diverticulum presenting as a blind pouch connected to the bladder; a urachal cyst containing a small patent lumen with two closed ends; and an umbilical-urachal sinus, which communicates with the umbilicus but not with the bladder (6). The most common anomaly is the urachal cyst, with an estimated incidence of approximately 1 in 5,000 births (7). Diagnosis may be challenging, as imaging findings can resemble other anomalies such as bladder diverticulum or incomplete bladder duplication (8). Postnatal imaging modalities including ultrasound and voiding cystourethrography may assist in establishing the diagnosis (6).

Foetal MRI may serve as the next step in the diagnostic approach of foetal abdominal cysts. Hügele et al. demonstrated that foetal MRI has superior diagnostic performance compared with ultrasound, increasing the diagnostic accuracy for foetal abdominal cystic lesions from 51% to 73.4% when added to

the assessment (9). The recommended diagnostic workup and differential diagnosis for foetuses presenting with isolated ascites are summarized in Table 3 (10–12). In our case, a thorough prenatal evaluation was performed to determine both the cause of the ascites and the aetiology of the cyst. Although foetal MRI has superior diagnostic performance in the evaluation of foetal abdominal cysts, it was not performed in this case due to the clear visualization of the lesion on ultrasonography and MRI was therefore considered unlikely to provide additional diagnostic value. In addition to imaging, genetic analysis testing was performed in this patient. Previous authors have stated that an isolated abdominal cyst is not, in itself, an indication for genetic testing, which should therefore be reserved for foetuses with additional structural abnormalities (4). In our case, parental carrier screening for cystic fibrosis was performed and yielded negative results. However, cystic fibrosis testing is not routinely indicated in the absence of sonographic features suggestive of meconium peritonitis, such as calcifications. Nevertheless, it may be considered given the relatively high carrier frequency in the Caucasian population.

Prenatal follow-up was primarily guided by the presence of both the foetal ascites and the cyst. Depending on the underlying aetiology, potential complications associated with a foetal abdominal cyst include bowel



FIGURE 2: Postnatal ultrasound of the abdominal cyst located anterocranial from the roof of the urinary bladder extending towards the umbilicus without clear connection with the bladder (measuring 1 cm AP x 1.6 cm LL x 2.3 cm CC)

obstruction (particularly if an intestinal cyst is detected), development of polyhydramnios and preterm labour. Therefore, serial ultrasound and follow-up is necessary (1). In addition, the presence of ascites also has implications on prenatal management. Current recommendations advise ultrasound follow-up at 1–2-week intervals to monitor foetal growth, progression of ascites and amniotic fluid volume (10). Current evidence on intrauterine treatment of ascites remains controversial. In the past, intrauterine paracentesis or abdominal-amniotic shunting was considered where foetal development was compromised due to the amount of ascites (10). However, due to the rapid reaccumulation of ascites, intrauterine paracentesis didn't prevent complications (13). In patients presenting with a combination of these clinical entities, thorough evaluation is essential to support effective collaboration among neonatologists, gynaecologists, and paediatric surgeons. This multidisciplinary approach is crucial to provide optimal parental counselling, as well as guiding obstetric and neonatal care (3,4).

Relationship between posterior urethral valves, foetal ascites and a urachal cyst

Antenatally there was already a suspicion of lower urinary tract obstruction. Due to the thickened bladder wall, the diagnosis of PUV was re-evaluated by performing repeat voiding cystourethrography on the seventh day. When PUV is suspected, voiding cystourethrography should be performed as soon as

possible to confirm the diagnosis (14). Voiding cystourethrography was able to confirm the presence of PUV despite the absence of hydronephrosis. Posterior urethral valves represent the most common cause of congenital obstructive uropathy (15). When urinary tract obstruction is suspected, antenatal evaluation should include assessment of amniotic fluid volume and detailed ultrasonographic examination, with the “keyhole sign” representing the classic sonographic hallmark. It is also possible to detect the presence of a urachal remnant by using voiding cystourethrography, especially when the urachal remnant presents as a diverticulum. However, when the remnant presents as a urachal cyst containing a small patent lumen with two closed ends, voiding cystourethrography can be insufficient to detect this remnant. In this patient, MRI of the abdomen was needed to confirm the diagnosis. Due to the presentation of ascites, a broad diagnostic approach was provided in our patient during admission on the neonatology ward. At present, there are no guidelines recommending the use of genetic analyses such as next generation sequencing to detect metabolic diseases in neonates presenting with ascites. However, it is currently standard clinical practice to perform a gene panel to investigate treatable metabolic diseases. This was performed due to the important therapeutic consequences and clinical outcome when a treatable metabolic cause is detected.

The coexistence of the following clinical entities (ascites, urachal cyst and posterior urethral valves) may

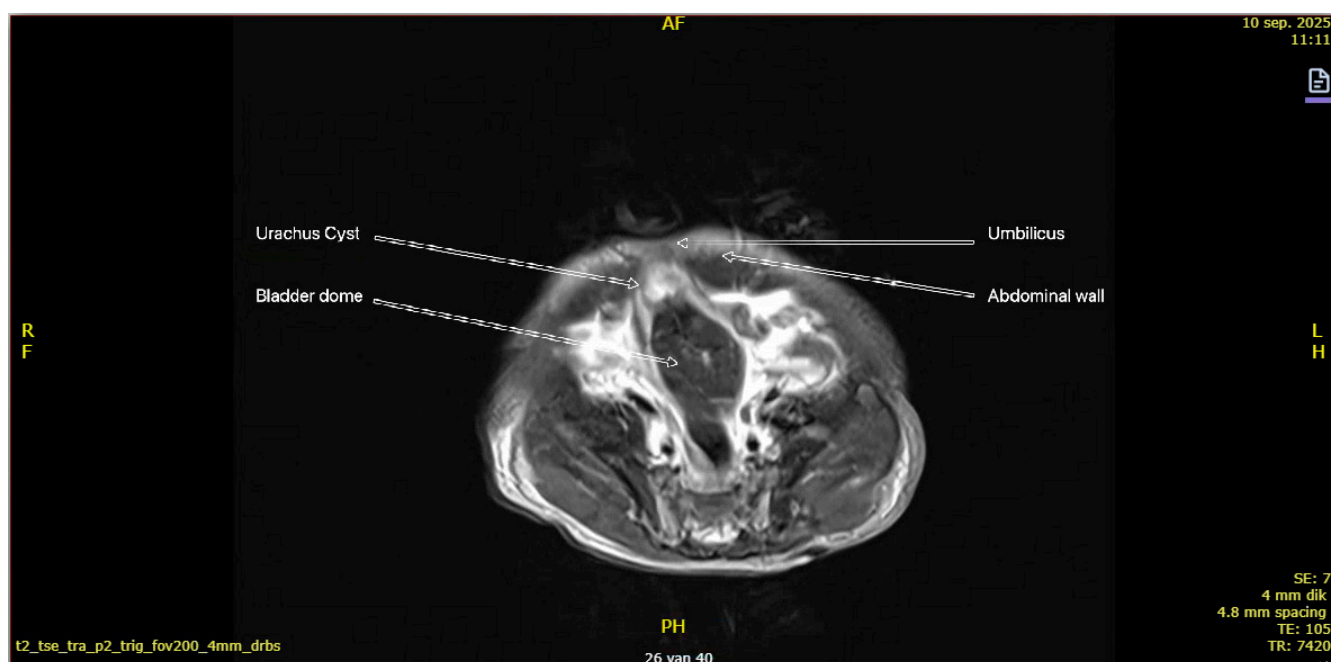
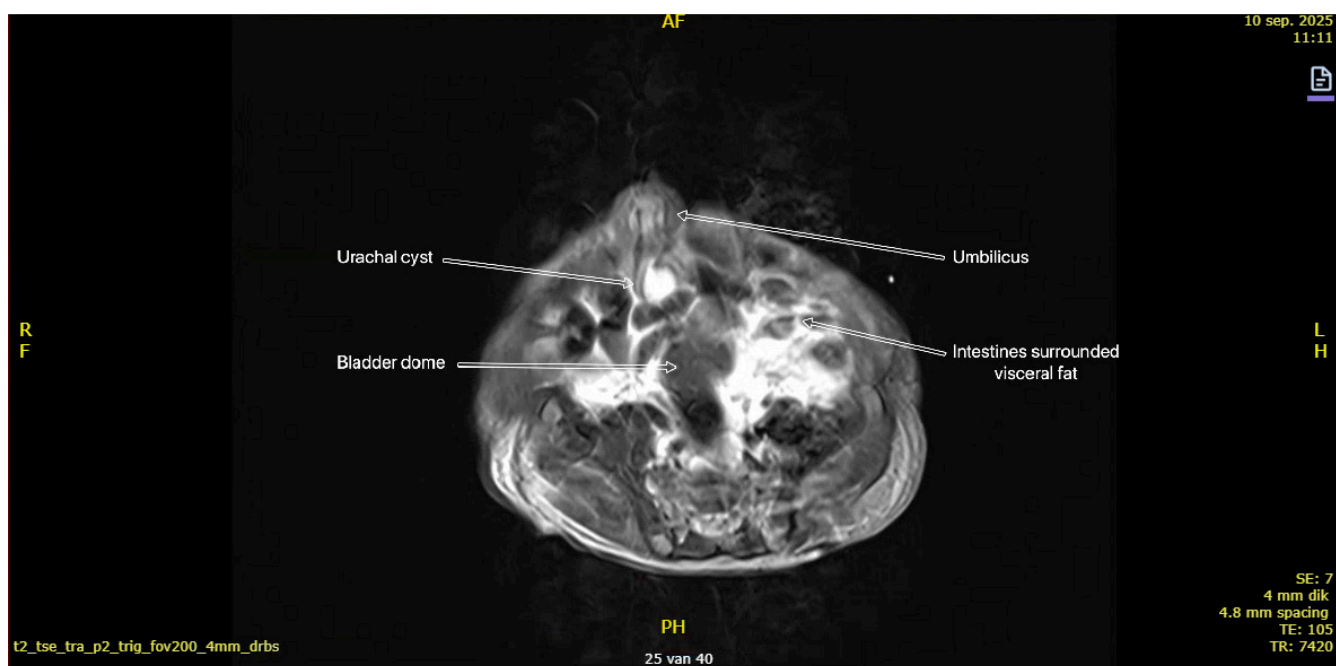


FIGURE 3a & b: MRI of the abdomen demonstrating the urachal cyst.



explain the unusual clinical presentation observed in this patient. There is a known association between PUV and the presence of a patent urachus (14). A case of a neonate with urinary ascites and posterior urethral valves without hydronephrosis was already reported by Prat et al. A hypothesized mechanism is the 'pop-off' mechanism (14–16). This mechanism is seen when increased pressure proximal to the obstruction could lead to forniceal or calyceal rupture, resulting in urinary ascites (which is life threatening when occurring in neonates) or a perinephric urinoma (in cases of extraperitoneal urinary extravasation). Several compensatory mechanisms have also been described, such as large bladder diverticula and urinary extravasation, which are known mechanisms to

buffer high pressures in the urinary tract in utero and may therefore preserve renal function (15). It is also plausible that the urachal cyst functioned as a pressure-relieving pathway, limiting the transmission of increased intravesical pressure to the upper urinary tract. In this scenario, pressure generated by the urethral obstruction may have decompressed through the urachal remnant rather than resulting in hydronephrosis. The urinary ascites may therefore have resulted from partial rupture of the cyst or from transudation of urine into the peritoneal cavity. If ascitic fluid consists predominantly of urine, elevated concentrations of creatinine and urea would typically be expected. However, the biochemical composition of urinary ascites may be altered by partial resorption of fluid

TABLE 2: Differential diagnosis of foetal abdominal cysts (1,3,4).

Organ system	Lesion	Frequency	Characteristics
Embryological malformation	Urachal cysts	Rare	
Endocrinological	Adrenal (haemorrhagic) cysts		
Genital	Ovarian cysts	Most frequently in female fetuses	Location to the side of the bladder
	Hydrometrocolpos		
Intestinal	Intestinal duplication cysts	Most frequent	
	Hepatic cysts	Frequent	
	Biliary cysts	Frequent	
	Meconium pseudocysts	Frequent	Calcifications
	Mesenteric cysts	Frequent	
	Splenic cysts	Frequent	
	Bowel dilatations	Frequent	
	Choledochal cyst		Relationship of the cyst to the hepatic artery
	Pancreatic cyst		
	Anorectal anomalies		Pelvic cysts
Oncological	Cystic tumours, such as neuroblastoma	Rare	
Pulmonary	Intra-abdominal extra lobar bronchopulmonary sequestration with cystic changes		
Urological	Renal cysts		

across the peritoneum. The peritoneal membrane can act as a dialysis membrane between urine and plasma (17,18). As urine contains higher osmolarity and higher concentrations of creatinine, urea, and potassium, whereas plasma contains higher sodium levels, an “autodialysis” phenomenon may occur, allowing diffusion of water and sodium into the peritoneal cavity (17,19,20). Consequently, creatinine levels in urinary ascites are generally lower than in urine but remain higher than serum creatinine levels (20). Nevertheless, it remains controversial that the ‘pop-off’ mechanism seen with urinary extravasation is indeed kidney protective. The systematic review conducted by Khondker et al. demonstrated that urinary extravasation was not statistically significant associated with a renal protective effect (16). Illustration 1 schematically illustrates the proposed pressure-relieving pathway that may have functioned as a pop-off mechanism.

Further management of our patient is guided by the guidelines on congenital lower urinary tract obstruction and the current evidence on the management of urachal anomalies. The guidelines of the European Association of Urology and European Society for Paediatric Urology state the importance of immediately postpartum management consisting of prompt bladder drainage and initiation of antibiotic prophylaxis, when lower urinary tract obstruction is suspected. Given the potentially life-threatening nature of this condition, close monitoring in a neonatal intensive care setting is required, including regular assessment of

renal function and electrolyte levels (14). Once the diagnosis is confirmed, definitive treatment consists of endoscopic valve ablation. Despite optimal management, approximately 20% of patients may progress to end-stage renal disease, highlighting the need for life-long monitoring (14).

Due to the rarity of urachal anomalies, evidence-based management guidelines remain limited. Urachal cysts are frequently asymptomatic but may occasionally lead to complications such as infection, peritonitis, enteric fistula formation, or, rarely, malignant transformation. Both benign and malignant tumours have been reported, including teratoma, mucinous tumours, giant cell tumours, rhabdomyosarcoma, and yolk sac tumours (8). Management options include surgical excision or conservative follow-up. Increasing evidence supports conservative management in asymptomatic paediatric patients, as spontaneous postnatal involution of urachal remnants has been described (5). Regular monitoring using ultrasound imaging is advised and has a high sensitivity (nearly 80%). Consequently, prophylactic excision during the first year of life is generally not recommended (8,21). A review by Ghattas et al. supports this conservative approach, with surgical intervention reserved for symptomatic cases, particularly in patients presenting with recurrent infections (8). When surgery is required, open excision remains the traditional gold standard, although laparoscopic and robotic approaches have also been reported to be safe and effective alternatives with improved

TABLE 3: Diagnostic workup and differential diagnosis in fetuses with isolated ascites (10–12)

Organ system	Differential diagnosis	Additional diagnostic technique
Cardiac	Heart failure (due to structural cardiac defects)	Structural ultrasound on foetal cardiac echography
	Cardiac Arrhythmias	
Chylous	Lymphatic malformations	
Gastro-intestinal	Meconium peritonitis	Genetics: Prenatal testing cystic fibrosis. Imaging: hyperechogenic bowel, dilated bowel and intraabdominal calcifications
	Small intestinal or colonic atresia	
	Intestinal perforation	
	Cloacal dysgenesis	
	Omphalocele	
	Oesophageal atresia	
	Volvulus	
	Intussusception	
	Biliary atresia	
	Portal venous malformations	
Genetic	Trisomy 18 or trisomy 21	Amniocentesis for karyotyping
	Turner syndrome	Amniocentesis for karyotyping
	Osteogenesis imperfecta type II	
	Others*	
Genital	Ovarian cyst	
	Persistent cloaca/cloacal dysgenesis	
Hematologic	Haemolytic disease of the neonate (Rh or ABO alloimmunization)	Maternal antibody screening and direct antiglobulin test
	Severe congenital anaemia	Kleihauer–Betke test; Doppler flow of the umbilical/cerebral arteries
Infectious	Cytomegalovirus, parvovirus B19, hepatitis A/B/C, herpes simplex 1, 2, varicella, rubella	Serology
	Syphilis, Listeriosis	Bacterial cultures and serology
	Toxoplasmosis	Serology
Metabolic	Niemann–Pick type C	
	Infantile Sialic Acid Storage Disease	
	Sialidosis	
	Mucopolysaccharidosis VII	
	Congenital disorders of glycosylation	
	Wolman disease	
	Lysosomal storage disease	
Obstetric	Severe foetal growth restriction	Abnormal umbilical artery dopplers
	Severe placental dysfunction	
Pulmonary	Congenital cystic adenomatoid malformation of the lung	
	Congenital hernia diaphragmatica	
	Laryngeal stenosis	
Urological-nephrological	Posterior urethral valves	
	Urogenital sinus anomaly	
	Polycystic kidney disease	
	Dysplastic kidneys	
	Lower urinary tract obstruction	
	Ureteropelvic junction obstruction	
	Vesicoureteral reflux	
	Prune belly syndrome	

*Others: cardiosplenic syndrome, Fraser syndrome, chromosome 4p deletion, Sotos syndrome, stomatocytosis, primary lymphangiectasia, skeletal dysplasia, McKusick Kaufman syndrome and 46, XXt(1;4) (q31; q34) (11).

cosmetic outcomes (6). In accordance with the current literature concerning management of urachal cysts (3,6), this patient didn't receive prophylactic surgery yet and currently is treated with prophylactic antibiotics (trimethoprim) due to low grade active vesicoureteral reflux. Currently a cystoscopy is planned at one year of age, and if the cyst persists, an excision will be performed due to the small but potential risk of malignancy.

Strengths and limitations

To our knowledge, no previous cases describing the coexistence of isolated foetal ascites, urachal cyst, and posterior urethral valves have been reported. This case underscores the importance of a thorough diagnostic evaluation in neonates presenting with unexplained ascites in combination with a urachal remnant. Furthermore, the present review provides an overview of the differential diagnosis of foetal abdominal cystic lesions and foetal ascites, which may assist clinicians when confronted with similar clinical presentations. It should be noted that the conditions summarized in the tables are derived from the performed narrative review and are therefore not exhaustive. Given the broad spectrum of clinical entities addressed, a narrative review methodology was considered the most appropriate approach. However, compared with a systematic review, this methodology has inherent limitations, including the absence of a formal quality assessment of the included studies,

thereby limiting the ability to formulate specific evidence-based recommendations.

Conclusion

Prenatal detection of an abdominal cyst in combination with isolated ascites requires a thorough diagnostic evaluation. Early recognition of this unique combination, particularly in the presence of bladder wall thickening, may raise suspicion for posterior urethral valves, and thereby facilitate timely diagnosis and appropriate postnatal management. A plausible explanation is that the urachal cyst functioned as a pressure-relieving mechanism, thereby accounting for the absence of hydronephrosis. Clinical follow-up will be necessary to determine whether this presumed pressure-relief mechanism translates into a more favourable long-term renal prognosis.

Statements

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Written informed consent for publication of the medical data and imaging studies was obtained from the patient's parents.

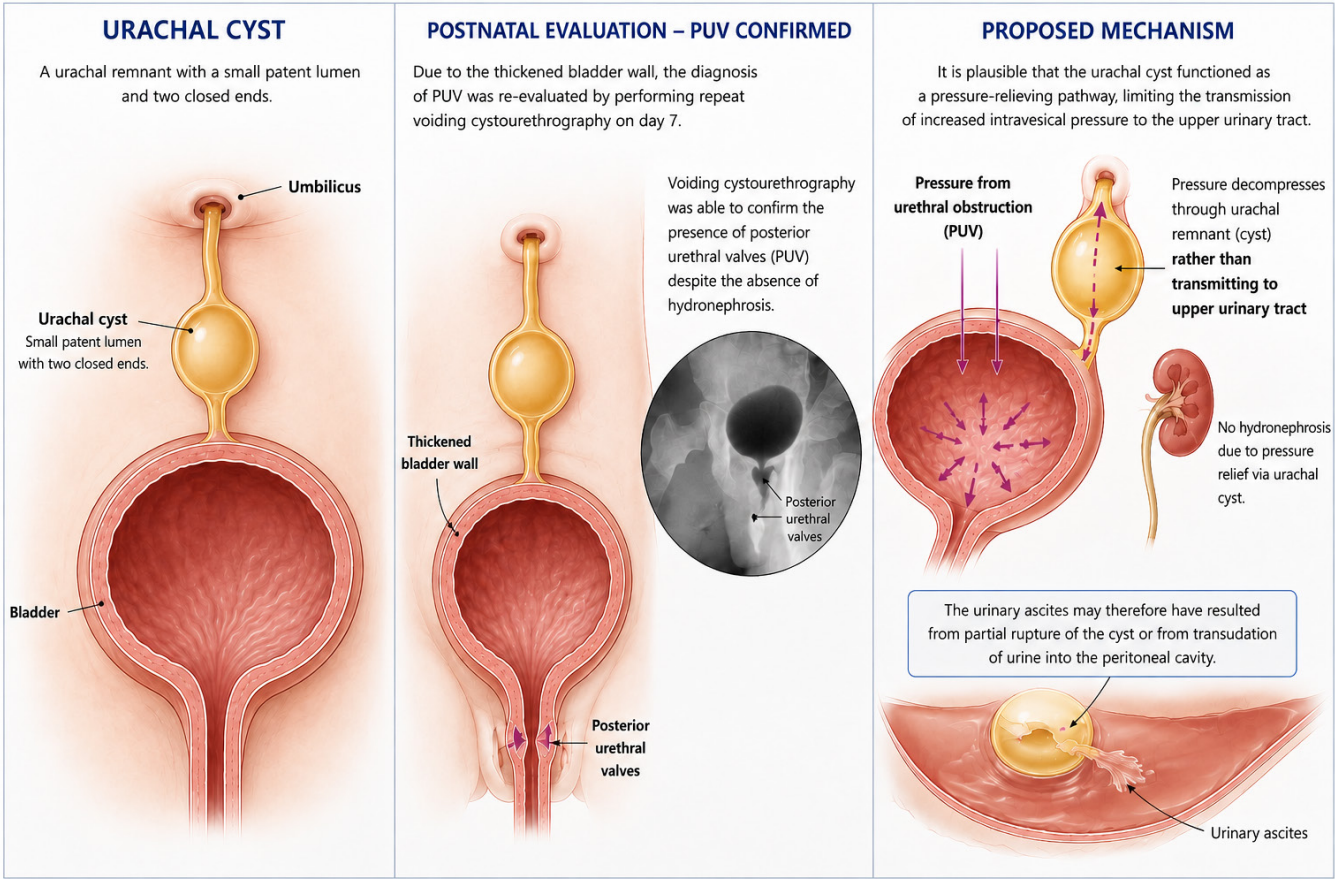


ILLUSTRATION 1: Urachal cyst and proposed mechanism of pressure-relieving pathway.

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