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## CASE REPORT

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# Rupture of Renal Pelvis Secondary to Obstructing Calculus in Menkes Disease: A Case Report

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## INTRODUCTION

Menkes disease is a rare X-linked hereditary multisystem disorder due to a defect in copper metabolism caused by a mutation in the *ATP7A* gene.<sup>1</sup> Patients can present with neurodegenerative manifestations and multiple connective tissue abnormalities.<sup>1</sup> We report the case of a 4-year-old boy, known to have Menkes disease, who presented with an obstructive ureteric stone complicated by rupture of the renal pelvis.

## CASE REPORT

Our patient was diagnosed with Menkes disease at 6 months of age. Since then, he has developed recurrent urinary tract infections, attributed to the presence of multiple urinary bladder diverticula. He has received copper histidine injections since around 8 months of age and clean intermittent catheterisation since 19 months of age.

The patient presented with an episode of abdominal distension and septicaemia at the age of 4. There was no history of recent surgery or trauma. Ultrasound

of the abdomen and pelvis revealed right-sided hydronephrosis and fluid collections around the right perinephric and right subphrenic regions (Figure 1). Further contrast-enhanced computed tomography (CT) scan of the abdomen and pelvis showed an obstructing stone at the right proximal ureter with gross upstream hydronephrosis. Delayed phase images were subsequently obtained at 60 minutes post injection and confirmed the presence of a mural defect at the posterior aspect of the right renal pelvis with contrast extravasation from the corresponding area and contrast pooling around the right perinephric region (Figure 2). A ruptured right renal pelvis was diagnosed.

Following diagnosis, insertion of a double-J stent was attempted for urinary diversion but was unsuccessful due to difficulty in identifying the ureteric orifice in the presence of multiple urinary diverticula. Percutaneous nephrostomy was performed instead the following day. Ultrasound-guided drainage of a retroperitoneal collection was also performed subsequently for control of sepsis.

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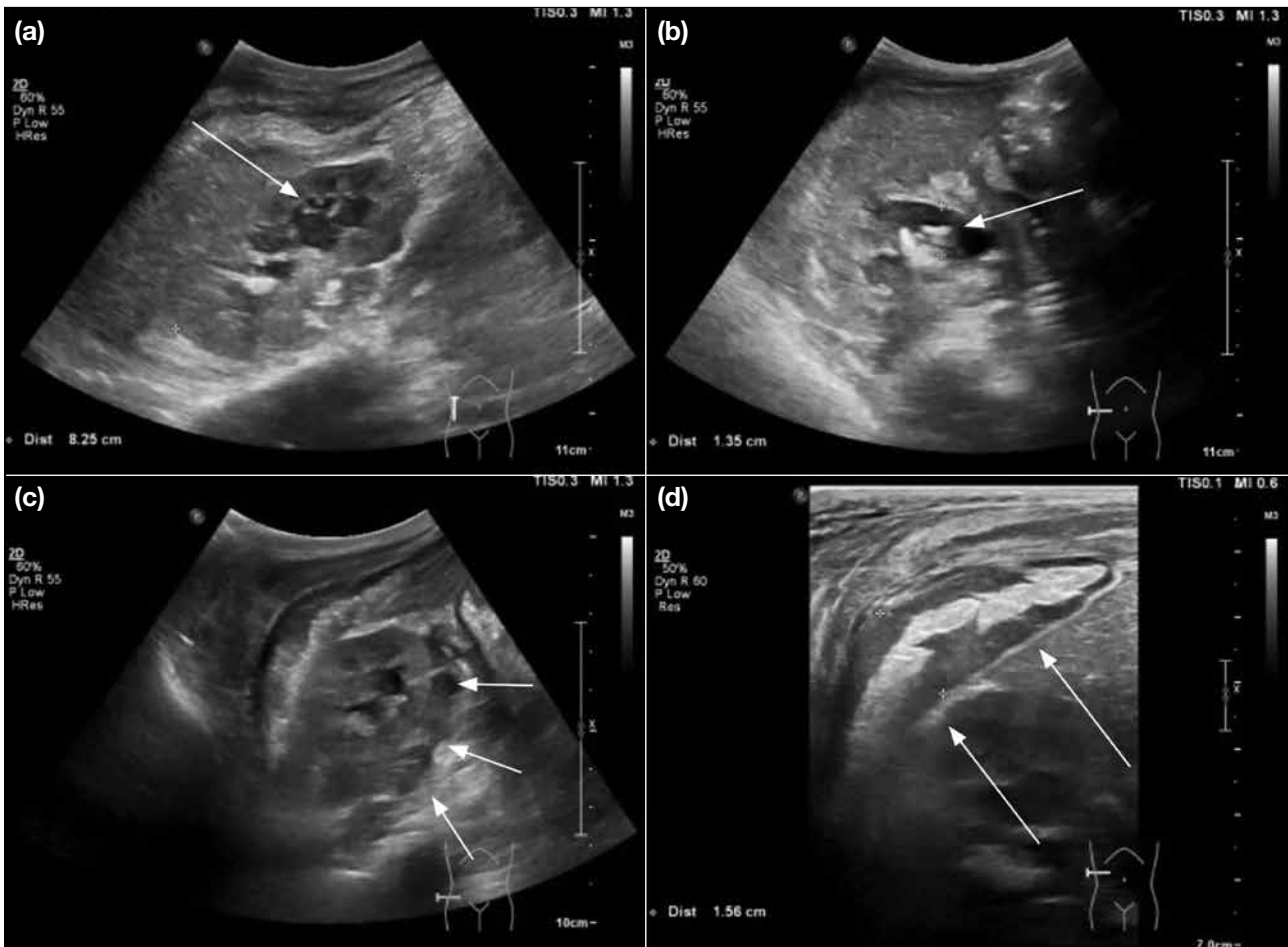
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**Figure 1.** Ultrasound images of the patient annotated with arrows showing (a) dilated calyces at the right kidney, (b) stones within dilated right renal pelvis, (c) perinephric collection at the posterolateral aspect of the right kidney, and (d) right subphrenic collection.

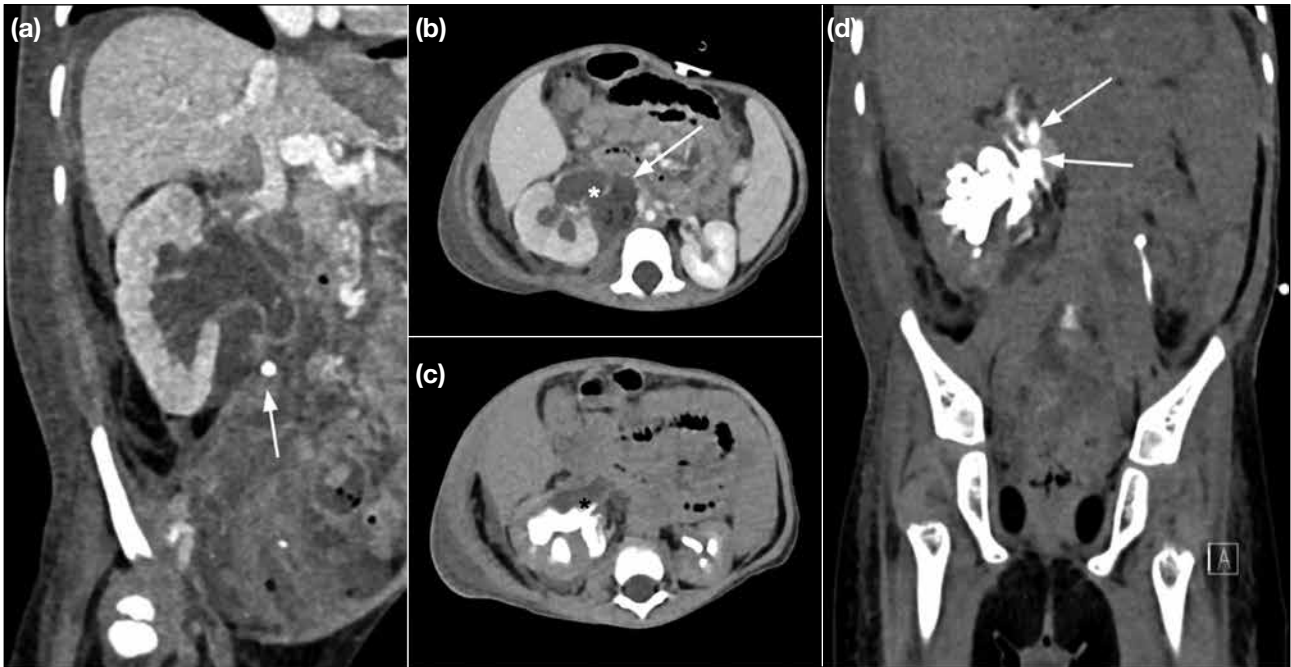
Open pyeloplasty was performed around 2 months after the first presentation as underlying ureteric stricture was suspected. Surgery was unsuccessful and complicated by persistent stricture. Further balloon dilatation was attempted but this also failed. After multidisciplinary discussion, a decision was made for a long-term internal ureteric stent to remain in situ. Our patient is now well following removal of the percutaneous nephrostomy catheter. A ureteric stent remains in situ and clean intermittent catheterisation continues.

## DISCUSSION

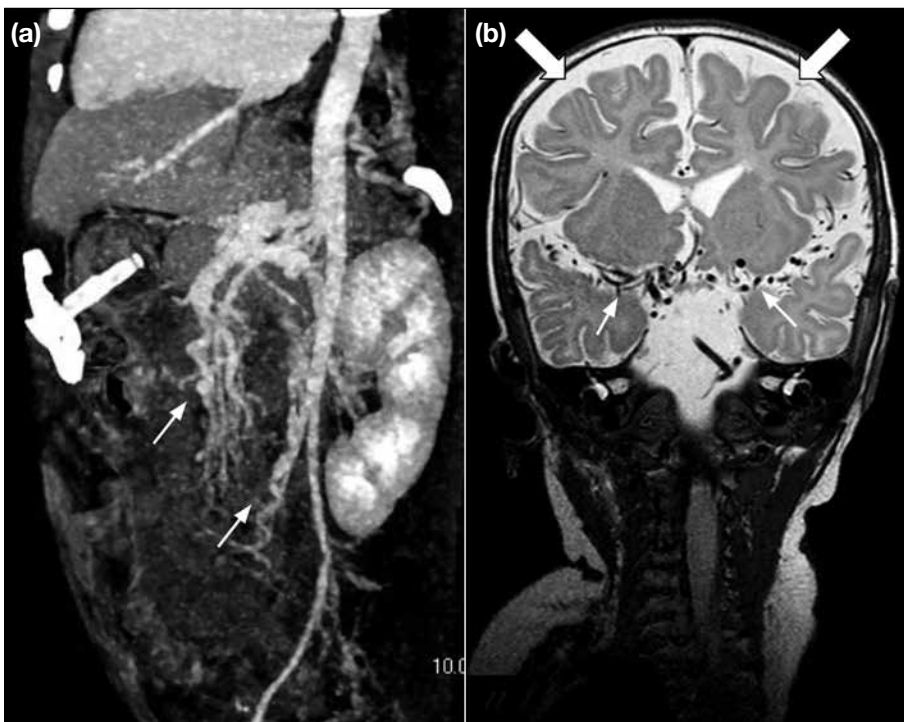
Menkes disease has an estimated incidence of 1 in 35 000 population<sup>2</sup> to 1 in 360 000 population<sup>1</sup>, varying widely in different localities. The *ATP7A* gene encodes a protein that is responsible for transcellular copper transport. A defect in such protein causes impaired

copper absorption in the intestines and consequent copper deficiency. This in turn results in reduced activity of various copper-dependent enzymes throughout the body, leading to neurodegeneration and defective connective tissue synthesis, manifesting in the form of connective tissue abnormalities such as frail hypopigmented hair and vascular tortuosity (Figure 3).<sup>1</sup>

Urological abnormalities are also frequent in Menkes disease. While the most common is urinary bladder diverticula (Figure 4), others such as vesicoureteric reflux, dilatation of the pelvicalyceal system and/or ureters, and rupture of urinary bladder diverticula have also been reported. These conditions may lead to urinary stasis and can predispose patients to recurrent urinary tract infections and urinary calculus formation.<sup>3</sup>



**Figure 2.** Contrast-enhanced computed tomography of the abdomen and pelvis of the patient. (a) Oblique coronal image showing an obstructing calculus at the right proximal ureter (arrow) with upstream hydronephrosis. (b) Axial image showing right hydronephrosis with focal mural defect (white asterisk) at the posterior aspect of the right renal pelvis and associated perinephric fluid collection (arrow) on axial images. (c) Delayed phase axial image showing contrast extravasation (black asterisk) from the renal pelvis. (d) Delayed phase coronal image showing contrast extravasation and pooling around the perinephric region (arrows).



**Figure 3.** (a) Three-dimensional reconstructed sagittal image from contrast-enhanced computed tomography of the abdomen and pelvis of the patient on maximum intensity projection setting showing tortuous intra-abdominal vessels (arrows). (b) Selected T2-weighted coronal image from magnetic resonance imaging of the brain obtained at 5 months of age showing tortuous intracranial arteries along the bilateral Sylvian fissures (thin arrows). Also note that there are diffusely prominent subarachnoid spaces (thick arrows), most likely due to reduced brain parenchymal volume.



**Figure 4.** Antegrade pyelogram of the patient in prone position. Contrast injection via right percutaneous nephrostomy catheter opacifies the dilated right pelvicalyceal system and shows a stricture along the right proximal ureter. Urinary bladder shows multilobulated contour (arrows), in keeping with multiple bladder diverticula.

Rupture of the urinary collecting system is rare in the paediatric population. It has been reported in the presence of trauma<sup>4</sup> or due to obstruction of the collecting system, for example by stone or congenital anomaly (e.g., posterior urethral valve or ureteropelvic junction obstruction).<sup>5,6</sup> Conditions that cause soft tissue abnormalities such as Klinefelter syndrome and Cushing's syndrome have been reported to be associated with ureteric rupture and are thought to be predisposing factors.<sup>7,8</sup> We believe that Menkes disease was likely a predisposing factor for renal pelvic rupture in our patient.

Clinically, it is difficult to differentiate renal pelvic rupture from other causes of abdominal pain and tenderness; hence, the mainstay of diagnosis is imaging. In the paediatric population, ultrasound is usually the first-line imaging investigation to determine an underlying cause of acute abdominal pain due to its lack of ionising radiation. Nonetheless it is difficult to confirm the diagnosis of renal pelvic rupture by ultrasound alone. In cases with a high index of suspicion, contrast-enhanced CT urography can help confirm the diagnosis, albeit at the expense of ionising radiation.

In our patient, the most suspicious feature on ultrasound was the presence of perinephric and subphrenic fluid collection. This warranted further imaging with contrast-enhanced CT to delineate its extent and cause. As review of subsequent CT images was suspicious of renal pelvic rupture, delayed phase images were obtained. The presence of contrast extravasation from the collecting system confirmed the diagnosis.

Treatment should be individualised for each patient and take account of the degree of sepsis, extent of urinoma or abscess formation, and any suggestion of persistent urinary leakage. Small urinomas can resorb over time and may not require drainage. In cases of sizable urinoma, image-guided drainage and urinary diversion in the form of percutaneous nephrostomy or double-J stent can be considered. Surgical repair of the defect may be appropriate for ongoing urine leakage.

## CONCLUSION

Rupture of the renal pelvis is an uncommon condition and difficult to diagnose clinically, particularly in children. Imaging with ultrasonography and contrast-enhanced CT urography can aid diagnosis and facilitate prompt management. Non-surgical treatment options are commonly considered for drainage of urinoma and urinary diversion, with surgical treatment reserved for cases of persistent urinary leakage.

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