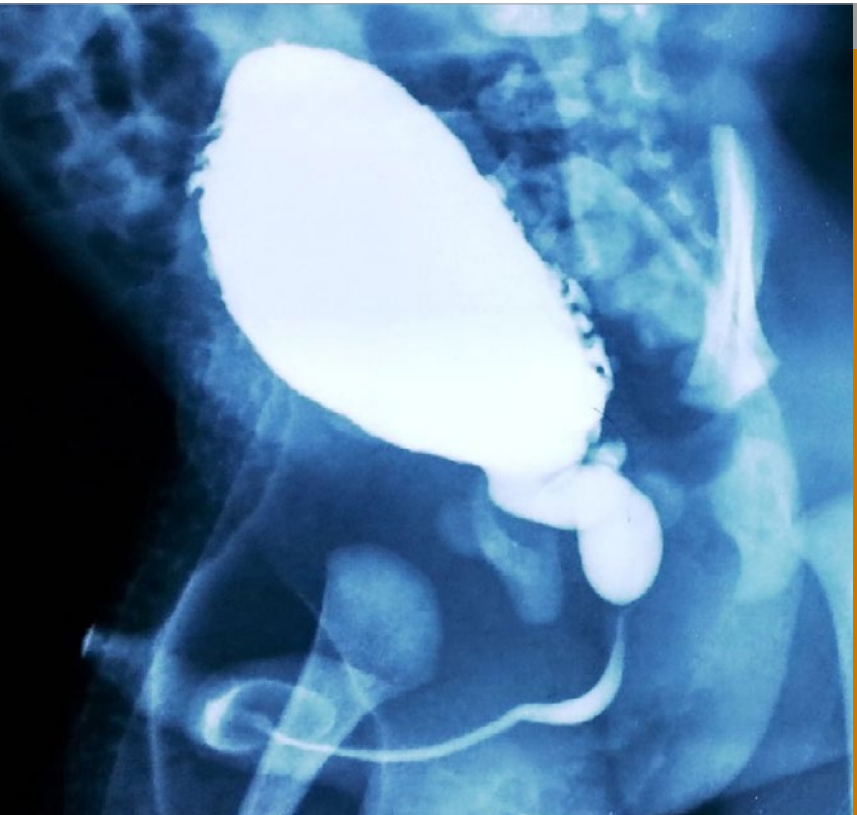
SWISS SOCIETY OF NEONATOLOGY

Atypical presentation of posterior urethral valves in an infant with Down syndrome



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Title figure:
MCUG: posterior urethral valves
(source: https://www.pediatricsurgeonindelhi.in)

INTRODUCTION

Posterior urethral valves are a frequent cause of urinary tract obstruction in infants and can lead to chronic renal disease. We report on a male newborn with an uncommon presentation of posterior urethral valves mimicking a bladder mass.

This male infant was born at 36 3/7 weeks of gestation to a healthy 33-year-old G2/P2 from Eritrea. The mother had just recently arrived in Switzerland seeking asylum. At 32 weeks of gestation, fetal ultrasound examination at our institution had been unremarkable, with no signs of malformations and normal amniotic fluid volumes. Previous antenatal visits had reportedly taken place in Sudan.

The mother was admitted to the labor ward with regular contractions. The baby boy was born by an uneventful, rapid vaginal birth and adapted well with Apgar scores of 9, 10, and 10 at 1, 5, and 10 minutes, respectively. Arterial umbilical cord pH value was 7.28. His birth weight was 2885 g (P45), length 48 cm (P25) and head circumference 35 cm (P75).

On initial physical examination, a number of clinical stigmata were noted: generalized muscular hypotonia, ocular hypertelorism with upslanting eye lids and medial epicanthal folds, a third fontanelle, a flat nasal bridge, macroglossia, a single transverse palmar crease on the left hand, as well as bilateral sandal-gap toes. These findings were felt to be consistent with Down syndrome.

A rapid test (QF-PCR) was positive for trisomy 21. Later on, the diagnosis was confirmed by conventional karyotyping (47, XY, +21). On echocardiography, a large atrial septal defect (ASD) with moderate left-to-right

shunting and a patent ductus arteriosus (PDA) were demonstrated. Hypothyroidism was excluded.

Initially, the boy stayed with his mother in the maternity ward. On the second day of life, he was transferred to the neonatal unit due to feeding difficulties, temperature instability, and gross hematuria. When voiding, the urinary stream seemed unimpaired. A full blood count revealed polycythemia and moderate thrombocytopenia.

An ultrasound of the abdomen was performed to exclude renal vein thrombosis. There were no signs of thrombosis, but an extremely thickened, irregular urinary bladder wall was noted; in addition, there was mild right-sided hydronephrosis (Fig. 1, 2). Various specialists agreed that this impressive finding most likely represented a neoplasia of the urinary bladder. Of note, during the ultrasound examination almost no urine was seen in the bladder.

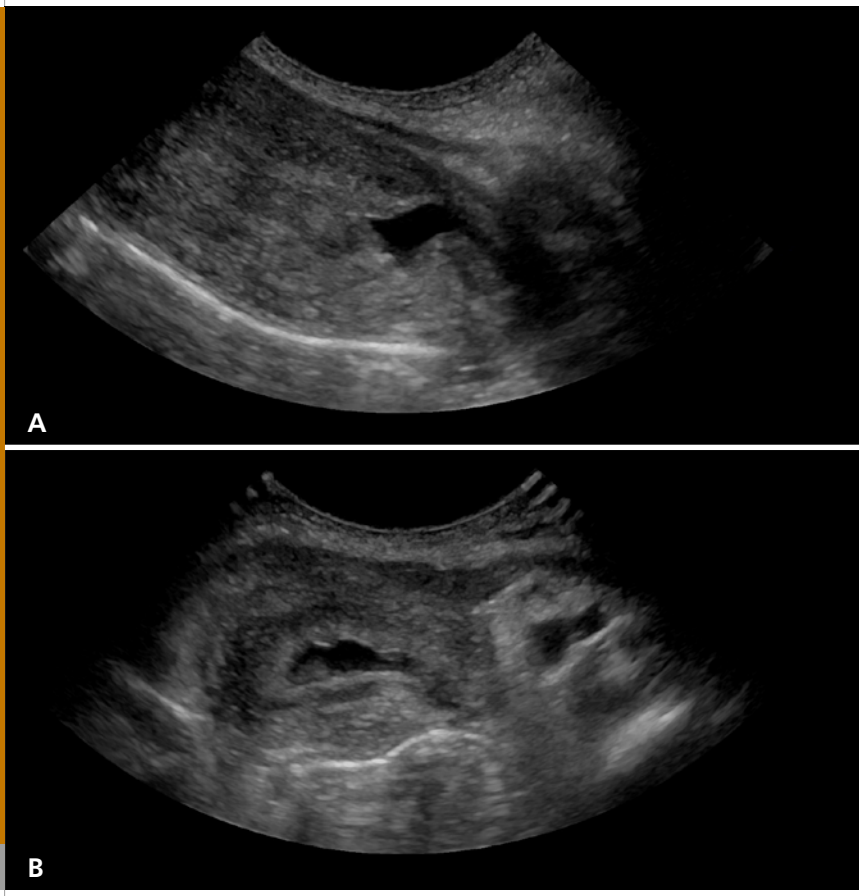

Fig. 1

Ultrasound examination of the urogenital tract on DOL 2: the urinary bladder is barely filled, and its wall appears irregular and thickened (A: sagittal view, B: transverse view).

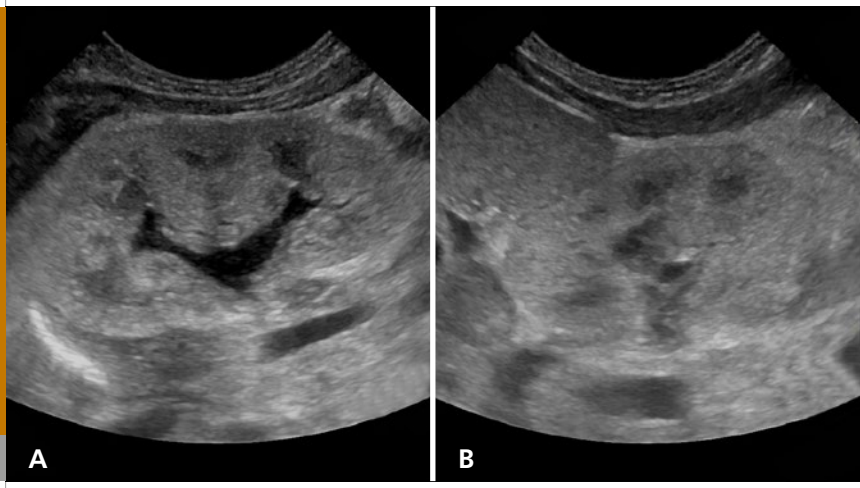

Fig. 2

Ultrasound of the urogenital tract on DOL 2: mild hydronephrosis of the right kidney (A), normal appearance of the left kidney (B).

On admission to the neonatal unit, serum electrolytes were normal, and urea and creatinine were only slightly elevated. Macrohematuria resolved spontaneously within 4 days, and all laboratory values normalized.

On regularly scheduled ultrasound examinations, mild bilateral hydronephrosis and dilatation of the ureters became increasingly evident, felt to be due to lower urinary tract obstruction. The bladder wall still appeared irregular and thickened, even though less impressively so than on the initial images (Fig. 3). At this point, antibiotic prophylaxis was started.

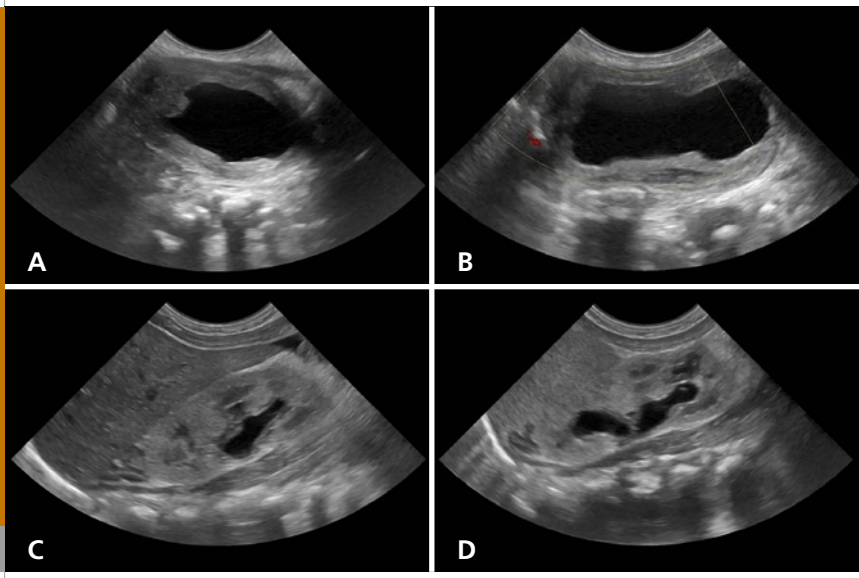

Fig. 3

Ultrasound examination of the urogenital tract on DOL 4: the bladder wall is still irregular and prominent, but less impressively so than on DOL 2 (A: sagittal plane; B transverse plane); mild bilateral hydronephrosis (C: right, D: left).

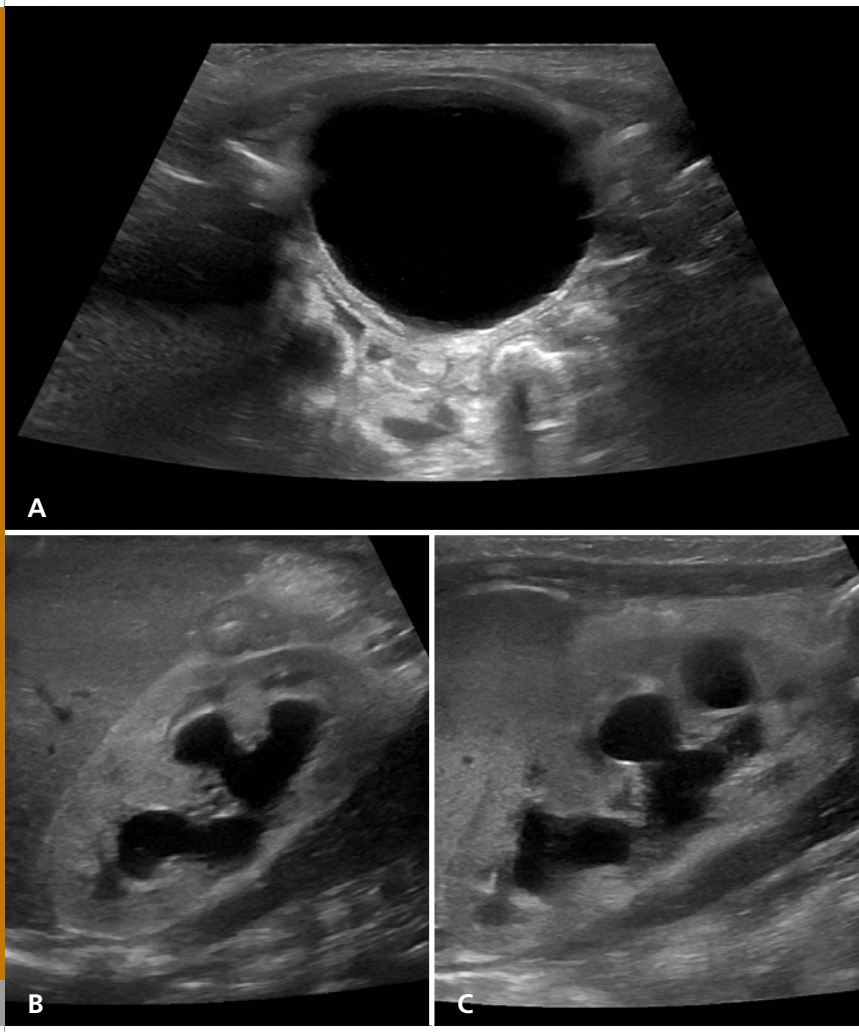

Fig. 4

Ultrasound examination of the urogenital tract on DOL 7: the bladder is well filled, and its wall appears slim (A); progressive bilateral hydronephrosis of both kidneys (B: right, C: left).

On day of life (DOL) 11, gross hematuria reoccurred, and both leukocyturia and bacteriuria were noted. Afebrile urinary tract infection was suspected, and intravenous antibiotics were administered. Urine culture showed growth of atypical bacteria, which were interpreted as contamination. Therefore, intravenous antibiotics were stopped, and oral antibiotic prophylaxis was resumed.

To further investigate the lower urinary tract obstruction, cystoscopy and voiding cystourethrogram (VCUG) were done on DOL 13. Cystoscopy revealed posterior urethral valves (PUV), and ablation of the valves was performed. Biopsies were taken from a part of the bladder wall that appeared irregular and tumorous. A VCUG showed no reflux (Fig. 5).

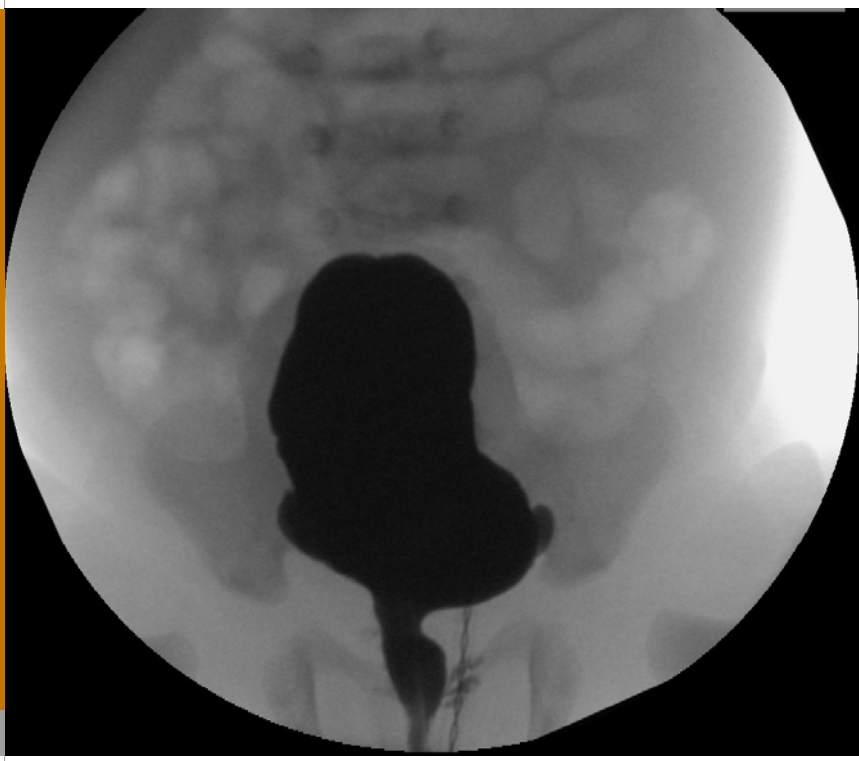

Fig. 5

Intraoperatively performed VCUG on DOL 13 showing no vesicourethral reflux, but an elongated irregularly formed bladder.

A transurethral splinting catheter that had been placed during surgery was removed on the 3rd postoperative day when slight reduction of hydronephrosis could be documented on follow-up ultrasound examination. Subsequently, micturition was unimpaired and gross hematuria did not reoccur.

Histology (including immunohistochemical analysis) of the biopsy specimen showed a fibroepithelial polyp with no signs of malignancy (Fig. 6). This finding was felt to be secondary to the chronically increased bladder pressure.

On day of life 20, the boy was transferred to a hospital closer to home for further care.

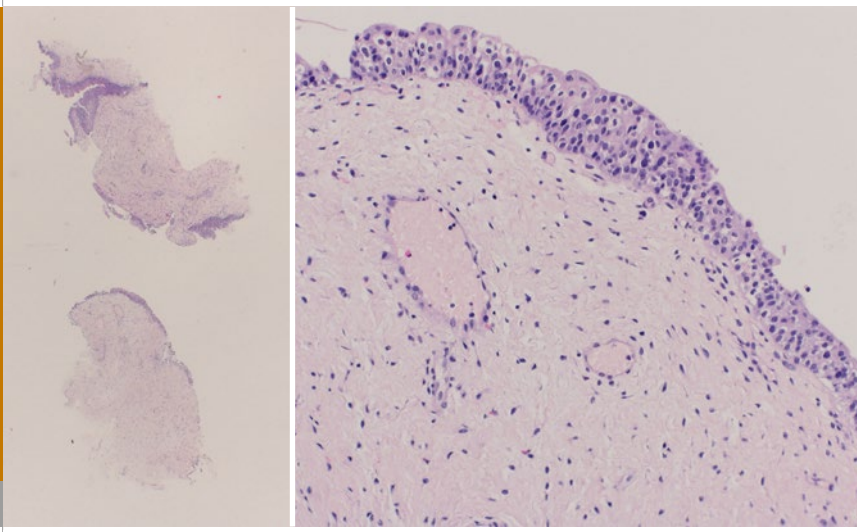

Fig. 6

Histology of bladder biopsies (A: overview, B: high power view): fibroepithelial polyp with minor signs of chronic inflammation, no signs of malignancy.

DISCUSSION

Posterior urethral valves (PUV) are congenital obstructing membranes within the lumen of the posterior urethra. They only occur in males (1). With an estimated incidence of 1:4'000 – 8'000, it is the most common cause of both urinary tract obstruction in male infants and chronic renal disease in children (1, 2).

Up to half of the patients with PUVs can be identified prenatally (3). In severe cases, PUV can lead to oligohydramnios, which can impair lung development and cause lung hypoplasia (4). Despite improvement in ultrasound technology, a large proportion of patients with PUV are detected only after birth (2). Patients may present with urinary tract infections, abdominal distension, voiding difficulty, or poor urinary stream (3, 5).

In our case, the patient presented with gross hematuria, a symptom not typically associated with PUV. Other symptoms like voiding difficulties were not noted. In addition, there had been no abnormalities (such as oligohydramnios) on prenatal visits. On an ultrasound examination on the second day of life, the urinary bladder wall appeared extremely thickened and irregular, raising the suspicion of a bladder tumor.

Bladder neoplasias are extremely rare in neonates. Rhabdomyosarcoma is the most common bladder tumor in children with an estimated incidence of 4 in 1'000'000 live births. The presenting symptoms of

bladder tumors may include hematuria, inflammation, or urinary obstruction (6, 7). There is no specific symptom or ultrasound finding for rhabdomyosarcoma, and benign or reactive bladder masses can mimic malignant lesions (6). The fibroepithelial polyp observed in our patient was felt to be secondary to the chronically increased bladder pressure.

Our case highlights the importance of an adequately filled bladder for ultrasound examination. During the initial ultrasound examination of our patient, the bladder was barely filled, enhancing the impression of thickening of the bladder wall. Even though this was noted, attending physicians from different subspecialties were still misled. The patient's signs and symptoms were thought to be explained by a bladder tumor and other underlying etiologies were initially not considered.

Of interest, patients with Down syndrome have an increased risk for renal and urinary tract anomalies (RUTAs) such as PUV. With a prevalence of about 3 %, RUTAs are about four to five times more frequent in infants with Down syndrome compared to the general population (8). However, routine screening for RUTAs in Down syndrome patients is not standardized.

CONCLUSION

An adequately filled bladder is essential for ultrasound examination of the urogenital tract. If the bladder wall of a neonate appears tumorous on ultrasound examination, the possibility of a bladder tumor must be considered. However, due to the rareness of bladder neoplasias in neonates, more common differential diagnosis should be ruled out first. In other words: when you hear hoof beats, think horses, not zebras!

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