Urology Case Reports 46 (2023) 102294

Contents lists available at ScienceDirect

Urology Case Reports

journal homepage: www.elsevier.com/locate/eucr

Urology case report: Multifactorial bladder dysfunction in the setting of down syndrome

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ARTICLE INFO

Keywords:
AKI
Bladder dysfunction
Down syndrome
Tarlov cyst

ABSTRACT

A 16-year-old male patient with Down syndrome diagnosed with AKI and urinary tract infection was treated with meropenem for ESBL-positive *E. coli* in urine culture. Persistently elevated creatinine and persistent post-void residual (PVR) of >300 mL led to further testing, which revealed urethral stricture and a lower sacral Tarlov cyst. Due to no complete improvement with urethral dilatation, he underwent laminectomy and Tarlov cyst fenestration. Creatinine normalized, with increased urine output and robust flow. Due to a PVR of >100 mL, he received behavioral therapy, including sitting and timed voiding, and the PVR was reduced to <5 mL.

1. Introduction

Children with Down syndrome have a higher prevalence of genitourinary complications, including bladder dysfunction, predisposing them to acute kidney injury. Traditionally, bladder dysfunction is attributed to either neurogenic, anatomic, or functional causes. However, literature that examines the multifactorial pathogenesis of bladder dysfunction in children with Down syndrome is lacking.

2. Case presentation

A 16-year-old male patient with Down syndrome presented to the ED at a referring hospital with abdominal pain, dysuria, and emesis for two days. He was prenatally and postnatally diagnosed with bilateral hydronephrosis, followed by nephrology every 6–12 months. At age 14, during follow-up periods, right and left kidney sizes were 15.2 cm and 17.1 cm, respectively (over 99th percentiles), serum creatinine of 0.6 mg/dL, post-void residual (PVR) of 335 mL (normal range for children <20 mL), with observed frequent holding maneuvers when voiding. After the first UTI (urinary tract infection) at age 15, he has had multiple episodes of UTIs with up-trending creatinine to 1.59 mg/dL. His parents endorsed a worsening inability to maintain a continuous urine stream and straining with voiding. A suprapubic catheter decreased creatinine to a nadir of 0.79 mg/dL, but subsequent worsening of creatinine and recurring UTIs were appreciated after the removal.

The patient was febrile at 38.6C at the ED, and lab results were significant for elevated leukocytes and nitrites in urinalysis, CRP of 306 mg/L, normal WBC, creatinine of 1.6 mg/dL, and cystatin C of 1.83 mg/dL. The pre-void volume and PVR were 318 and 249 mL, respectively. The treatment with ceftriaxone was switched to meropenem when the urine culture demonstrated ESBL-positive *E. coli*. Despite resolving fever and chief complaints, creatinine continued to rise to 2.0 mg/dL on day 4. He was then transferred to our hospital for a second opinion.

After a 7-day course of meropenem, we switched to oral ciprofloxacin, at which time creatinine was 1.4 mg/dL. Ultrasound and KUB demonstrated worsening bilateral hydronephrosis, right 19.3 cm and left 21 cm in length from his baseline (Fig. 1, Fig. 2). Bladder scans found continuously high PVR of over 300 mL. Given the consistent urinary retention, a spinal MRI was performed, with a significant result for a lower sacral 1.4 cm-sized Tarlov cyst, lateralizing the lower sacral nerve roots within the sacral canal (Fig. 3). Tarlov cysts are sacs filled with cerebrospinal fluid commonly found at the sacral level of the spine incidentally, which rarely causes compression of the spinal cord or nerve roots.

Additionally, a meatal urethral skin bridge was discovered during clean intermittent catheter teaching, preventing catheterization, and the patient required meatoplasty. He is circumcised but otherwise never had urethral inserting instruments previously. On cystoscopy during the procedure, narrowing the urethra distal to the bulbar urethra was incidentally found. In addition to the bridge correction, we completed urethral dilation for stricture on day 13. Although PVR decreased slightly above 100 mL with stabilized creatinine around 1.3 mg/dL after the procedure, we could not rule out neurogenic bladder secondary to a sacral cyst. He underwent S1-2 laminectomy and fenestration of

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https://doi.org/10.1016/j.eucr.2022.102294
Received 31 October 2022; Received in revised form 28 November 2022; Accepted 30 November 2022
Available online 6 December 2022

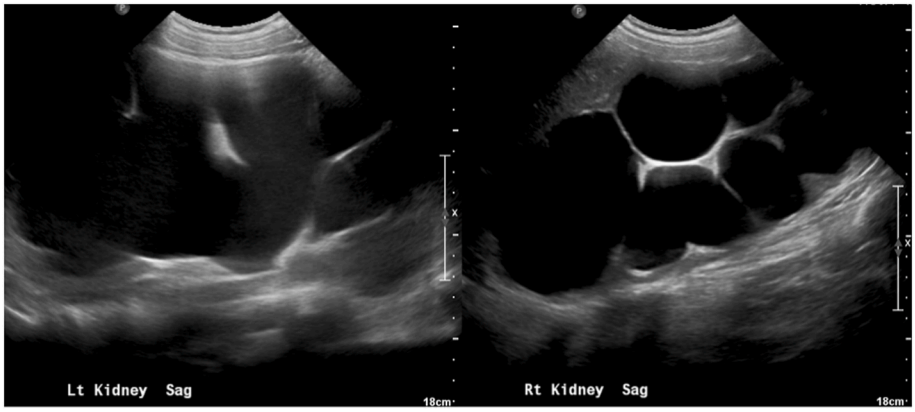

Fig. 1. Bilateral hydronephrosis (Ultrasound, right kidney on right side, left kidney on left side).

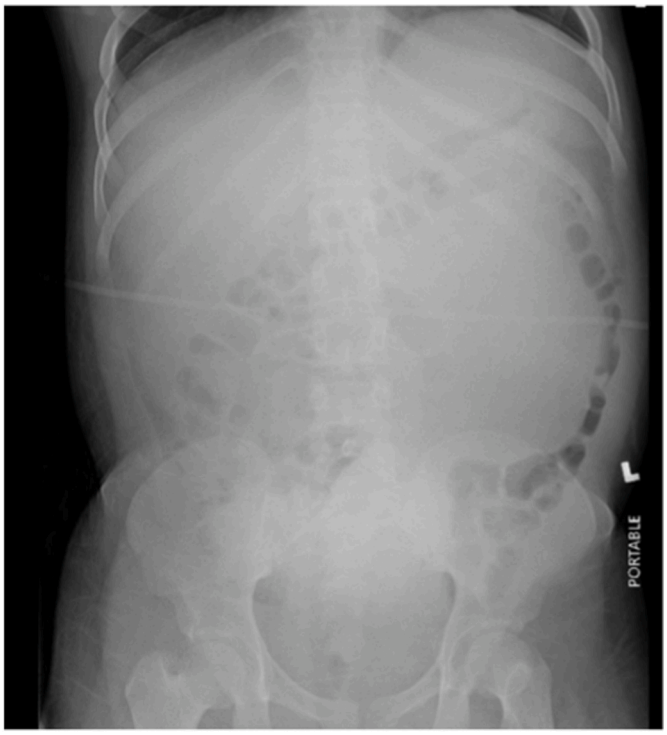

Fig. 2. Bilateral hydronephrosis (KUB).

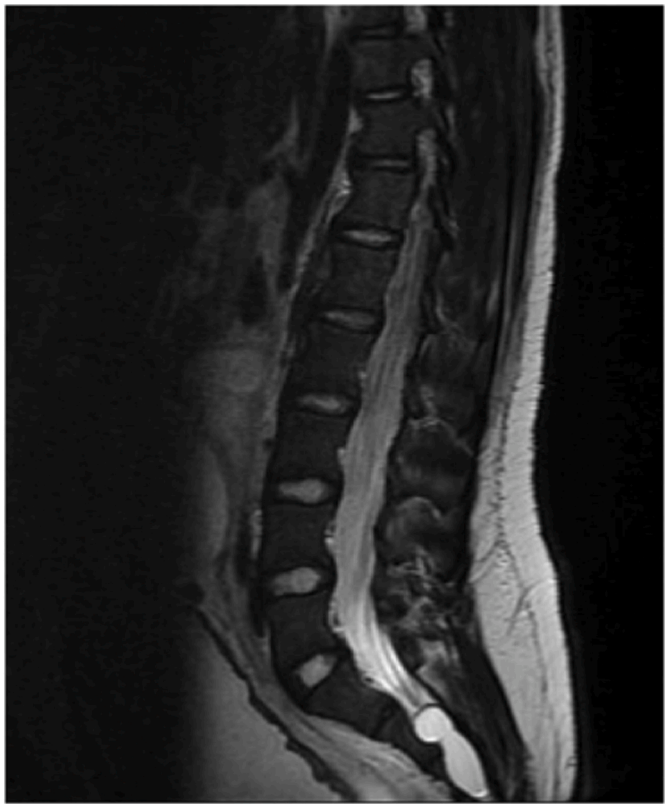

Fig. 3. Sacral tarlov cyst, 1.4cm (MRI T2).

intradural Tarlov cyst with the placement of a lumbar drain on day 22, which was complicated by cerebrospinal fluid leakage from the drain exit site after removal, being treated with a single suture.

The PVR remained around 100 mL, but creatinine dropped to 0.86 mg/dL. Parents also endorsed much higher urine volume with a more robust stream and spontaneous void without pushing. After discharge, he received behavioral therapy, including a timed voiding schedule with a sitting position while urinating. The length of bilateral hydronephrosis has decreased (right and left kidney size of 14.8 cm and 16.4 cm). A follow-up voiding study 7 months after discharge showed a PVR of only 2 mL.

3. Discussion

This is a case of recurrent UTI with multifactorial bladder dysfunction of neurogenic, anatomic, and functional causes. Bladder dysfunction is one of the risk factors for UTI in children.[1]

The pathology of Bladder dysfunction is categorized into three groups: Neurogenic, Anatomic, and Functional.[2] Neurogenic causes arise from damage to the central nervous system, including congenital anomalies, cord tethering, and spine or brain injury. Anatomic causes include ectopic ureter and urinary outlet obstruction like urethral stricture. Functional causes are known as behavioral or idiopathic. In this case, his chronic urinary retention due to over 300 mL of the PVR over a year had been thought to be functional, given the history of urinary holding maneuvers and straining behaviors, common complaints of children with Down syndrome.[3]

One challenge in patients with developmental disabilities is difficulty in diagnostic evaluation. Our patient exemplifies this challenge, with intolerance to procedures which resulted in delays in collecting objective data about his urinary retention through VCUG and urodynamics. We expected that the dilation of urethral stricture would resolve urinary retention, but with only a mild improvement. A Tarlov cyst presented a

diagnostic challenge, as these lesions are commonly found but rarely symptomatic. However, after the cyst fenestration, his creatinine dropped, and urinary frequency and strength of the urine stream improved significantly. In MRI imaging studies, the incidence of sacral perineural cysts is approximately 1.5–4.6%.[4] Most cases are asymptomatic, but 1% of the population with Tarlov cysts may demonstrate symptoms associated with nerve root compression. A study concluded that patients with Tarlov cysts >1.5 cm and with associated radicular pain or bowel/bladder dysfunction might benefit from surgical procedures.[5]

Interestingly, this patient's PVR of urine remained at 100–300 mL after the procedure and gradually decreased in outpatient follow-up with behavioral management. Minimal residual volume was observed after months of multidisciplinary management, which suggested that non-neurogenic and non-anatomic factors existed to some extent, either as a primary contributor or developing secondarily with his chronic retention and infections.

4. Conclusions

This case demonstrates an intertwined combination of neurogenic, anatomic, and functional causes; all components contributed to the overall clinical picture in this case. It shows the importance of a multidisciplinary approach for diagnosis and treatment with close follow-up in patients, especially with Down syndrome who present with acute or chronic genitourinary symptoms.

Funding

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Acknowledgments

Not applicable.

Abbreviations

AKI	Acute kidney injury
KUB	Kidney, urine, and bladder X-ray
PVR	Post-void residual
UTI	Urinary tract infection

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