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Case Report

Treatment of bilateral encrusted ureteral stents, large volume renal stones, and ureteral stricture in a complex pediatric patient: a surgical video and case review

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Abstract: Myotonic dystrophy is a debilitating genetic disease that carries a predilection for a variety of comorbidities. Kidney stone disease in this population can present a variety of unique challenges related to patient age, comorbidities, and social factors. We present a video review case of a 13-year-old girl with myotonic dystrophy who was treated surgically for large bilateral stone burden, bilateral retained ureteral stents with nephrostomy tubes, and right ureteral stricture. The patient had multiple prior urologic procedures and recurrent admissions for infection prior to presentation. Preoperative planning included non-contrast CT imaging, admission to an intensive care unit, and multidisciplinary discussion of treatment and goals. Through combined antegrade and retrograde approaches, the patient's stone burden was cleared, right ureteral stricture was treated, and all tubes were able to be removed in two major procedures and one minor cystoscopy with stent removal under anesthesia. Early referral to tertiary care centers and involvement of multiple specialist teams may help reduce perioperative risk and minimize the number of surgeries. Additionally, patients at high anesthesia risk may benefit from concurrent percutaneous nephrolithotomy with endopyelotomy.

Keywords: Myotonic dystrophy, percutaneous nephrolithotomy, nephrolithiasis, endourology, pediatric urology, video

Introduction

Myotonic dystrophy type 1 (DM1) is an autosomal dominant neuromuscular disorder that can affect multiple organ systems. Striated muscle in every organ as well as smooth muscle of the gastrointestinal tract are affected. Patients may appear normal at birth, however, can develop characteristic facial features such as an inverted-V-shaped upper lip and facial muscle wasting which can lead clinicians toward the diagnosis. As opposed to myotonic dystrophy type 2 (DM2) which involves a separate chromosomal abnormality and rarely presents in childhood, DM1 can present at any age, although typically it will manifest by adolescence and progressively worsen. Characteristic clinical features that may develop include muscle wasting in a distal distribution, weakness, myotonia (slow relaxation after muscle contraction), poorly articulated speech due to involvement of facial muscles, slow gastric emptying, cardiac arrhythmias, endocrine abnormalities, immune deficiency, cataracts, and intellectual impairment. In severe congenital forms of DM1, patients may exhibit symptoms immediately at birth and are at particular risk for respiratory compromise which may require ventilatory support. Characteristic laboratory findings may show normal or mildly elevated serum CK, hypothyroidism, and low immunoglobulin G. EKG may reveal evidence of heart block. The diagnosis of DM1 is made through DNA testing which reveals abnormal expansion of the CTG or CCTG repeat on chromosome 19. There is no treatment specifically to address DM1, and care is directed at managing the manifestations of the disease through the use of physiotherapy, cardiac surveillance, and respiratory support. Likewise, prognosis is dependent on the clinical expression of the disease and is highly variable [1].

Although myotonic dystrophy is known to affect multiple organ systems, its relation to the urinary tract, the smooth muscle of bladder detru-

sor, and urolithiasis is not well described in the literature and there is no clear link between them. Despite this, myotonic dystrophy patients have reported symptoms of infrequent voiding, stress incontinence, urgency, and urge incontinence. Various voiding patterns such as intermittent flow, high voided volumes, low flow rate, low urethral closure pressure, small bladder capacity, detrusor hyperreflexia, and bladder atony have been seen during urodynamic evaluation [2, 3]. Additionally, one case report has described the occurrence of cystinuria in brothers with DM1 and large renal stones, although the causal effect is unknown and this is not a commonly reported occurrence. Although prevalence studies describing the frequency of urolithiasis development amongst patients with DM1 is insufficient, it is clear that the sequelae of myotonic dystrophy may place patients at risk for development of urolithiasis. Urinary stasis and infection from voiding dysfunction and constipation, dysregulated calcium metabolism, and polypharmacy from comorbidities are important risk factors that can predispose these patients to develop stones [2, 4, 5].

The concurrence of myotonic dystrophy and nephrolithiasis can be devastating to both the patient and family members. Recurrent admissions for infections as well as the management of implanted tubes such as ureteral stents or nephrostomy tubes may present challenging social obstacles. The decision to proceed with surgical removal of stones must be balanced with the risks of anesthesia, particularly respiratory distress, and hypersensitivity to anesthetic agents [6, 7].

We report the case of a 13-year-old girl who had progressively worsening symptoms of myotonic dystrophy since birth, a longstanding history of nephrolithiasis and recurrent admissions for infection as well as repeated urologic interventions. We have included corresponding de-identified video clips from her surgery for review. With careful preoperative planning and involvement of multiple specialists, the patient was able to have bilateral large volume kidney stones removed, retained bilateral encrusted stents extracted, a right ureteral stricture treated, and all urologic tubes removed. In presenting this case report, we hope to bring attention to the importance of early referral to tertiary care centers and multidisciplinary specialist involvement to improve care for similarly complex patients. Additionally, our report highlights the importance of minimizing anesthesia exposure and preferential use of minimally invasive techniques such as concurrent PCNL with endopyelotomy in high-risk patients.

Case report

The patient is a 13-year-old girl with a history of myotonic dystrophy, nephrolithiasis, and recurrent infections. She developed sequelae related to DM since birth which have progressively worsened, namely intellectual disability, gastric tube dependence, severe scoliosis, hip dysplasia requiring surgery, and restrictive lung disease requiring home BiPap. She initially was able to feed herself until age 7 to 9 years old when she developed worsening dysphagia. Her complete loss of speech coincided with the onset of her recurrent infections, and nephrolithiasis developed around the same time period. Her mother elected not to have formal genetic testing for the patient, however, she is presumed to have DM1 due to the early onset of symptoms.

During this most recent admission, the patient presented to an outside hospital with sepsis attributed to urinary source and persistent desaturation requiring intubation for four days. Her full urologic history was not available for review, however the details were discussed with her pediatric urologist. The patient previously had recurrent nephrolithiasis and infections requiring multiple prior urologic procedures. The patient had bilateral retained ureteral stents from two years prior for which attempts at removal were unsuccessful due to encrustation. She also had a history of a prior right sided PCNL with known development of right ureteral stricture and recent unsuccessful extracorporeal shock wave lithotripsy (ESWL). The patient subsequently underwent placement of bilateral nephrostomy tubes, was treated for sepsis, and stabilized before being transferred to our hospital for definitive treatment of her nephrolithiasis, retained ureteral stents, and ureteral stricture.

Prior to transfer, as well as immediately on arrival, the patient had involvement of multiple specialist teams including Pediatric Intensive Care, Endourology, Pediatric Urology, Pediatric Nephrology, Infectious Disease, Anesthesia, and Social Work. Preoperatively, these teams

worked together to optimize the patient and plan for surgical intervention. A CT scan of the abdomen and pelvis was obtained which confirmed the presence of large bilateral stone burden, encrusted appearing ureteral stents, and indwelling nephrostomy tubes in appropriate position for urinary drainage, though poor position for percutaneous renal surgery. In speaking with the patient's parents prior to the procedure, it was revealed that care of the patient's nephrostomy tubes was a significant burden and that prompt and safe removal of the nephrostomy tubes was a key goal of care. Therefore, a major goal for our initial procedure was to leave the patient without nephrostomy tubes, and to ultimately leave her stone-free and stent-free through subsequent procedures.

Video surgery review (Supplementary Video x:xx designates the corresponding time in the surgical video)

Operation 1

On the day of surgery the patient had induction of general anesthesia and was carefully placed into the modified supine position for left percutaneous nephrolithotomy.

Cystoscopy, right ureteral stent removal: Cystoscopy was performed, and the right sided ureteral stent was noted to be internally encrusted. However, it was able to be removed relatively easily using a cystoscopic grasper. A retrograde pyelogram (**Figure 1A**) revealed hydroureter and two narrowings in the proximal ureter with complete obstruction at the level of the more proximal narrowing suggestive of a severe stricture. An antegrade pyelogram (**Figure 1B**) performed through the right nephrostomy tube showed severe hydronephrosis with no contrast passing down the ureter.

Right ureteroscopy confirming stricture: Retrograde diagnostic right ureteroscopy was performed. The more distal of the two narrowings was encountered ureteroscopically and was approximately 8 Fr in size and easily traversed. Of note, adjacent to this first area narrowing, stones were noted adjacent to the ureter and within the retroperitoneum on fluoroscopy. These likely represent stones avulsed from the collecting system during the patient's prior stone procedures and suggested the strictures may be related to inflammation secondary to retained stone fragments. Indeed, the more proximal narrowing was visualized to be a pinpoint stricture with stone shards embedded in the wall (Supplementary Video, 0:05). Initial attempts at passing both sensor and glide wires across the stricture were unsuccessful, however eventually the lumen was cannulated with a sensor wire (Supplementary Video, 0:17) and advanced fully with the assistance of a 5 Fr open-ended ureteral catheter. This was exchanged for a super stiff wire (**Figure 1C**) and a 6 Fr firm ureteral stent was placed in retrograde fashion (**Figure 1D**).

Cystoscopy, attempted left ureteral stent removal: Cystoscopy was then repeated and the distal curl of the left ureteral stent appeared severely encrusted. The distal end of the stent was able to be grasped, uncurled, and brought out of the meatus, however significant resistance was felt. An antegrade pyelogram was performed through the left nephrostomy tube to define the renal anatomy which revealed severe hydronephrosis with visible stone, and fluoroscopy confirmed that the stent was not unfurling proximally (**Figure 1E**). The distal end of the curl was clamped outside the body.

Left percutaneous access: The existing nephrostomy tube was felt to be a poor angle for stone clearance, so new access with 24 Fr sheath was obtained into the lower pole under ultrasonic and fluoroscopic guidance.

Lithotomy: A lithotripter device was then used to debulk the severely calcified proximal curl of the retained stent (Supplementary Video, 0:35). The nephroscopic grasper was used to straighten the stent (Supplementary Video, 0:44) which was able to be easily removed transurethrally. Additional stone was removed from the lower and upper pole calyces (Supplementary Video, 0:58).

Stone surveillance: Partial antegrade ureteroscopy was performed as the scope was unable to pass beyond the proximal ureter, however the visualized ureter was free of stone and antegrade ureterogram revealed a patent left ureter (Supplementary Video, 1:06; **Figure 1F**). Full flexible pyeloscopy was performed which revealed no further stones. Fluoroscopy likewise revealed no evidence of residual stone. A soft ureteral stent was placed.

Procedure finish: Repeat fluoroscopy of the right side prior to conclusion of the procedure

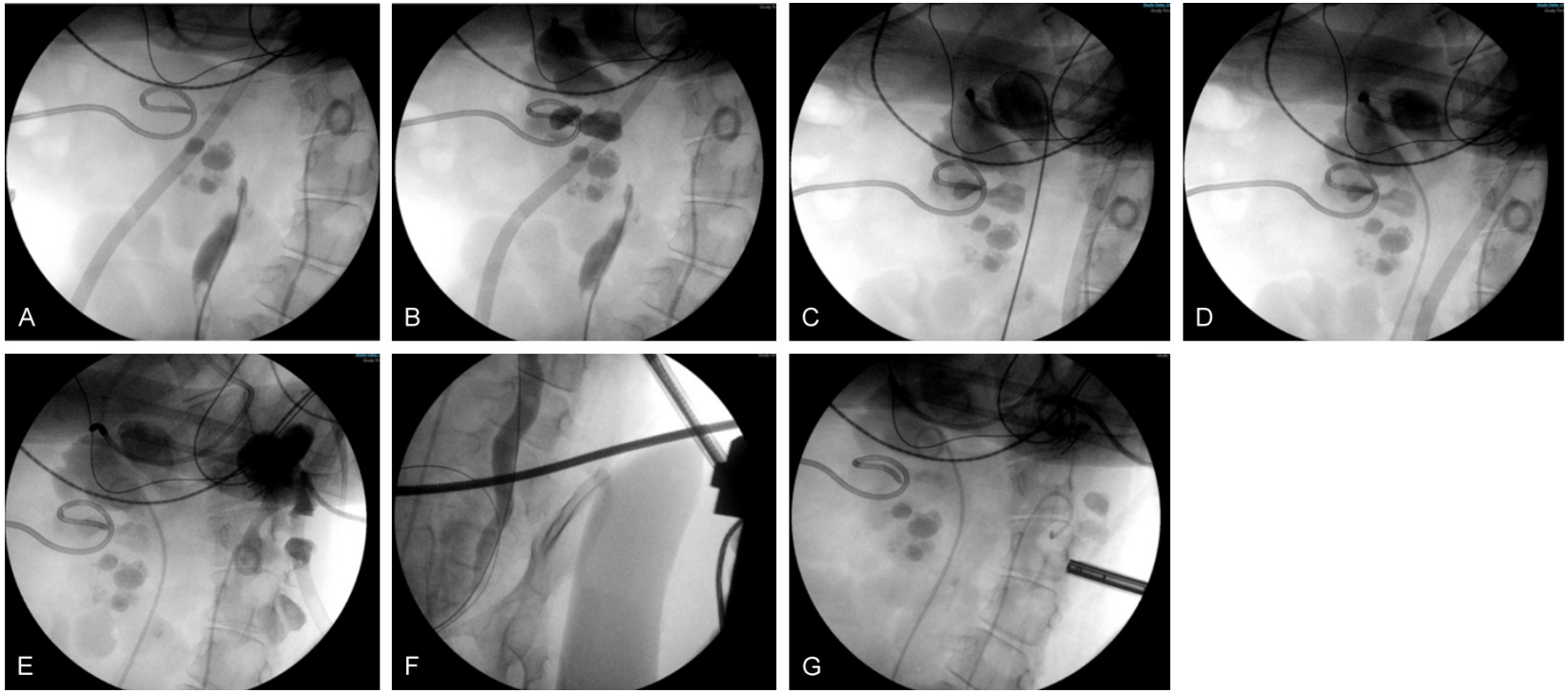

Figure 1. A. Right retrograde pyelogram revealing hydroureter and two narrowings in the proximal ureter with complete obstruction suggestive of stricture. B. Antegrade pyelogram performed through the right nephrostomy tube revealed severe hydronephrosis with no contrast passing down the ureter. C. A super stiff wire was able to be placed through an open ended ureteral stent. D. Placement of a firm right ureteral stent. E. Antegrade pyelogram revealing severe hydronephrosis. Gentle traction during removal of retained left ureteral stent confirmed that the stent would not unfurl due to encrustation. F. Left antegrade ureterogram revealing no evidence of ureteral stones. G. Fluoroscopy post left percutaneous nephrolithotomy revealing no evidence of residual stone. Left ureteral stent in position, and washout of prior contrast.

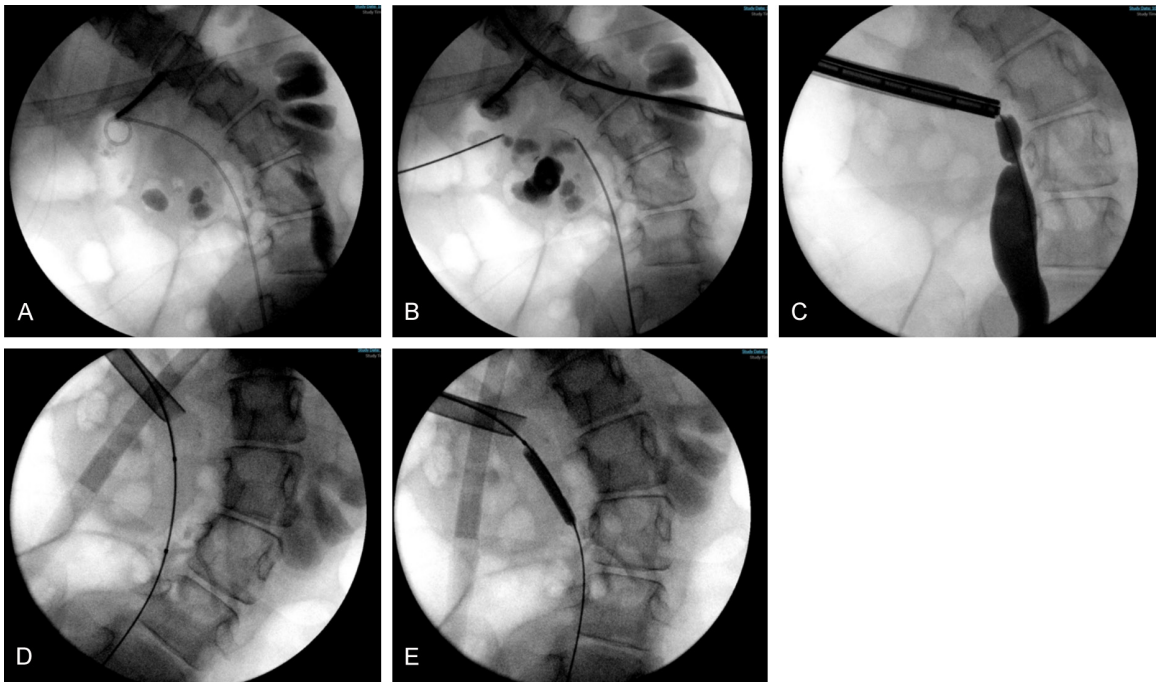

Figure 2. A. Left retrograde pyelogram following left ureteral stent removal that revealed residual left hydronephrosis, otherwise normal with no evidence of residual stone. B. Right retrograde pyelogram showed moderate right hydronephrosis and opacifications likely representing known large stone burden. C. Right antegrade and retrograde contrast instillation revealed a short stricture length, approximately 5 mm. D. An 18 Fr balloon was placed endoscopically across the stricture site and fluoroscopy confirmed the radioopaque markers to be in good position. E. The 18 Fr balloon was inflated with contrast to perform soft dilation of the ureteral stricture.

showed good washout of contrast (**Figure 1G**), therefore the right nephrostomy tube was capped to be removed in coming days and the left nephrostomy tube was removed.

On POD#1, the right nephrostomy tube was removed, rendering the patient free of her nephrostomy tubes, in line with goals of care.

Operation 2

Several weeks later the patient was brought back to the operating room and after induction of anesthesia was placed in the prone position.

Left ureteral stent removal: Cystoscopy was performed and the left ureteral stent was grasped and removed. Ureteroscopy was attempted, however the ureter was too small to cannulate. A left retrograde pyelogram was performed and revealed residual hydronephrosis but was otherwise normal (**Figure 2A**).

Right ureteral stent removal, retrograde pyelogram: The distal curl of the right ureteral stent was then grasped, a wire was placed easily into the right renal pelvis, and the right ureteral stent was removed. A retrograde pyelogram was performed showing moderate right hydronephrosis and opacifications likely representing the known large stone burden (**Figure 2B**).

Right percutaneous access: Right percutaneous access with a 24 Fr access sheath was obtained into the upper pole under the guidance of ultrasound and fluoroscopy.

Lithotomy: Significant clot and stone burden was cleared using a nephroscopic grasper and lithotripter device (Supplementary Video, 1:27). Flexible pyeloscopy confirmed no further significant stones seen within the renal collecting system (Supplementary Video, 2:18). The ureteropelvic junction (UPJ) was cannulated in antegrade fashion and noted to be tight around the open-ended ureteral catheter with a pinpoint lumen (Supplementary Video, 2:39).

Endopyelotomy: Contrast was instilled antegrade to define the length of the stricture and was estimated to be approximately <5 mm in length (**Figure 2C**). Laparoscopic shears were introduced via the nephroscope and used to

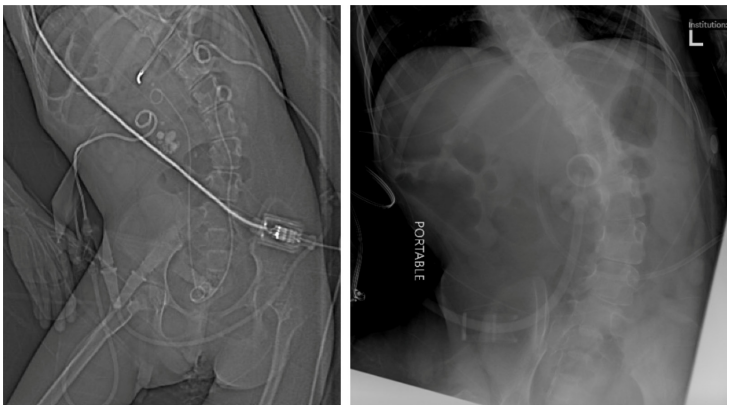

Figure 3. Representative preoperative CT scout imaging (left) shows large bilateral stones, bilateral encrusted ureteral stents, and bilateral nephrostomy tubes. Postoperative KUB (right) shows clearance of stone burden and removal of all urologic tubes.

perform a cold incision of the stricture extending the true lumen laterally (Supplementary Video, 2:57). Small stone shards were seen to extrude from within the stricture. All fibrous bands were incised and the UPJ was open and easily traversable with the nephroscope.

Triamcinolone injection, balloon dilation: 3 cc of triamcinolone was injected into the stricture bed in an attempt to prevent recurrence. An 18 Fr balloon dilator was then passed across the stricture site (**Figure 2D**) and soft dilation was performed under fluoroscopic guidance (**Figure 2E**) to imbue the triamcinolone within the stricture bed (Supplementary Video, 3:21). The balloon was noted to insufflate easily across the stricture bed with soft dilation suggesting complete treatment of the stricture.

Procedure finish: An 8 Fr right ureteral JJ stent was then placed (Supplementary Video, 3:32). Fluoroscopy was repeated for the left kidney with good washout of contrast. No nephrostomy tube was placed and the procedure was concluded. The patient's ureteral stent was removed one month later. Postoperatively the patient's stone analysis results revealed 100% calcium phosphate composition from the first surgery and 95% calcium phosphate with 5% calcium oxalate monohydrate from the second surgery. The patient's 24-hour-urine analysis was remarkable for low urinary citrate (164 mg/d) and alkaline urine pH (8.55).

Results

Postoperative CT scan on hospital day 22 prior to right stent removal revealed complete resolution of the right-sided stone burden and trace stone debris remaining in the left kidney, stable hydronephrosis, and possible small interval decrease in hydronephrosis despite the left ureteral stent being removed, consistent with chronic obstruction. On hospital day 43 RBUS showed no sizable stones and persistent chronic hydronephrosis with no worsening in hydronephrosis (and possible mild improvement) compared to prior to bilateral stents being removed. This was expected given her chronic renal obstruction since at least 2014. On hospital day 45 the patient's right ureteral stent was removed in the operating room. **Figure 3** shows preoperative CT scout imaging and postoperative KUB imaging demonstrating complete resolution of stone burden and removal of all urologic tubes. As of 2 months from stone clearance, the patient has remained UTI free and renal function has remained stable.

Discussion

The patient presented here has a history of myotonic dystrophy, which is a degenerative neuromuscular disorder that can affect multiple organ systems with variable severity [1]. The diagnosis and management of urolithiasis in this population is left to the discretion of the Urologist or treating clinician. There are no established guidelines to our knowledge, and clinical judgment is important to make inferences from the information at hand. The connection between myotonic dystrophy and the genitourinary system, including urolithiasis, has not been well reported in the literature, and there is much still to be investigated. There are, however, pathologic states in myotonic dystrophy which alter homeostasis and may lead to stone formation. Our patient was found to have a poorly optimized respiratory status leading to chronic acidosis, vitamin C deficiency from malnutrition, reliance on intermittent gastric tube feeding, and recurrent urinary tract infec-

tions. This is not uncommon among patients with myotonic dystrophy. As discussed, many patients have numerous medical issues including respiratory acidosis from hypoventilation and gastrointestinal abnormalities leading to alterations in absorption. Some patients cannot feed themselves and rely entirely on tube feeds which have variable formulations. Our patient's stone composition was almost exclusively calcium phosphate, and 24-hour urine testing revealed alkaline urine pH with hypocitraturia. The complex nature of myotonic dystrophy and the degree to which it affects many different organ systems makes it very challenging to determine the major driving factors leading to stone formation. While the etiology of these urine abnormalities is not entirely clear, and possibly multifactorial, we believe obtaining 24-hour-urine testing is especially crucial in these patients to guide preventative therapies.

Patients with myotonic dystrophy and concomitant nephrolithiasis can present with a wide array of symptoms and challenges that requires careful planning and discussion to overcome. This patient had progressively worsening symptoms since birth including severe restrictive lung disease requiring home BiPap, scoliosis, gastric tube dependence, and intellectual disability with inability to speak. In such patients, who cannot verbalize symptoms, it is important to consider further diagnostic testing such as CT imaging if concerning findings such as recurrent urinary tract infections, hematuria, or acute kidney injury are present. In addition to these chronic issues, the patient had an overwhelming number of urologic issues to address including recurrent urinary tract infections, right ureteral stricture, significant nephrolithiasis bilaterally, bilateral retained ureteral stents, and bilateral nephrostomy tubes which were difficult for her family to manage. Thus, the challenge presented was how to accomplish these treatments safely and in as few surgeries as possible.

The immediate involvement of multiple specialty teams to guide care for the patient was of utmost priority. A multidisciplinary approach to care has been defined as "a range of healthcare professionals working independently, but in a coordinated manner to improve outcomes, especially for chronic diseases". In pediatric patients with complex diseases such as this one, it is imperative that all aspects of the disease are addressed, as individual specialists or general practitioners may only see one 'piece of the puzzle'. Similarly, the utilization of a family-centered approach for children with partnership and shared decision making between parents and healthcare providers can improve effectiveness of care [8]. Our team worked diligently to engage all necessary specialists in perioperative planning and appreciate the goals and opinions of the parents.

Although myotonic dystrophy has not been found to specifically affect the pelvic floor or urinary sphincter, it has been connected to various forms of voiding dysfunction including infrequent voiding, urgency, and incontinence [2, 3, 9]. One study from the UK examining a large primary care database compared patients with neuromuscular disease (NMD) to age-sex-practice matched patients without NMD and found a nearly two-fold prevalence rate for hospitalization due to infection. Patients with myotonic disorders had the highest number of comorbidities compared to other NMDs, and myotonic dystrophy (type 1) had a 1.86 prevalence ratio (CI 1.20, 2.89) for genitourinary infections compared to matched patients without NMD [4]. As discussed earlier, the exact relationship between myotonic dystrophy and urolithiasis is not perfectly understood, however it is clear that risk factors such as voiding dysfunction, recurrent infections, polypharmacy, and altered metabolism put the patients at high risk for stone formation [2, 4, 5].

The decision to proceed with surgery in this patient was complex. Patients with myotonic dystrophy are at high risk for anesthetic complications. The reduced respiratory function of patients with myotonic dystrophy limits anesthetic management and such patients also frequently exhibit hypersensitivity to anesthetic agents. Additionally, there is risk for cardiac and pulmonary complications in the recovery period, particularly postoperative apnea and prolonged ventilator wean [7, 10]. The appropriate choice of anesthetic medications to avoid myotonic crisis and close monitoring of patients by experienced anesthesiologists, however, can lead to successful outcomes. Ultimately the decision to proceed was made based on the parents' goals and the assurance that specialist teams would provide excellent perioperative optimization.

In taking the above into consideration and through meticulous preoperative planning by our Endourology team, the patient was able to have all urologic issues addressed in two major procedure and one minor cystoscopy with stent removal.

Percutaneous endopyelotomy has been previously described, however, a generally accepted clinical principle is to perform endopyelotomy in a staged fashion from stone treatment, as precipitation of stone shards/stone debris within the stricture bed can promote stricture recurrence. Indeed, this patient's stricture was likely caused from residual stone fragments within the ureter from her prior procedures given that stone shards were found within the stricture at the time of endopyelotomy. However, given the extremely unique circumstances of this patient's case, and the challenges of general anesthesia in a patient with myotonic dystrophy, particularly with concurrent scoliosis and restrictive lung disease, we felt compelled to try and consolidate her care into as few procedures as possible. Though further follow up is required, the patient's stable (and possibly mildly improved) hydronephrosis, stable renal function, and lack of further UTIs, suggests that concurrent PCNL and endopyelotomy is a viable treatment alternative as long as a thorough and complete PCNL with complete and full treatment of the patient's stone burden is performed.

Conclusion

Myotonic dystrophy is a complex and incompletely understood disease with wide ranging clinical manifestations. Pediatric patients with myotonic dystrophy and urolithiasis necessitating surgical intervention are at high risk for complications and often require recurrent procedures. Referral of these patients to tertiary centers for specialized interdisciplinary care may provide the best opportunity for successful treatment. In such patients, in whom each anesthesia exposure is high risk, concurrent percutaneous nephrolithotomy with endopyelotomy can be considered.

Disclosure of conflict of interest

None.

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