Case Report

Hirschsprung's disease in a child with posterior urethral valve: An unwanted association

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ABSTRACT

Urinary bladder and the rectum share a common embryological origin, and the anatomical proximity of these two organs suggest that a dysfunction in either may influence the function of the other. Although, the coexistence of bladder and bowel dysfunction has been previously reported in the literature, there are hardly any reports on coexistence of posterior urethral valve (PUV) with Hirschsprung's disease. Here, we report a case of a 20-month-old male child who was initially treated for PUV and was later found to have coexisting Hirschsprung's disease.

INTRODUCTION

Posterior urethral valve (PUV) is the most common cause of bladder outlet obstruction (BOO) in male infants and may lead to urinary tract infections, urinary incontinence, chronic kidney disease (CKD), and even death.[1] The ill effects of PUV on the kidneys and the bladder may persist despite successful surgical management of the obstructing valves. Hirschsprung's disease, characterised by the absence of ganglion cells in the distal bowel, is a known pathological cause of intractable constipation in the early childhood. It has been suggested that the distension of rectum by impacted stool in constipated children may press on the bladder wall and can cause BOO or induce detrusor overactivity.[2] The association of pathological chronic constipation with lower urinary tract obstruction (LUTO) is rare and poses immense management challenge. Only few studies have described the association between congenital anomaly of kidney and urinary tract (CAKUT) and Hirschsprung's disease.[3] PUV in itself carries a significant risk of progressive renal disease despite timely intervention; chronic constipation caused by Hirschsprung's disease further adds to progressive bladder dysfunction and end-stage renal disease (ESRD).

CASE REPORT

A 20-month-old male child was admitted to the pediatric emergency with a 15-day history of fever, poor urinary stream, and constipation. On general physical examination, the child was febrile, lethargic, severely malnourished and had bilateral percutaneous nephrostomies (PCN) *in situ*. His weight for age was <3rd centile and height for age was found to be <10th centile of the Indian Academy of Pediatrics growth standards. The child was born through normal vaginal delivery and had uneventful antenatal and postnatal periods. He had a past history of poor urinary stream and constipation (one bowel movement every 2-3 days) as noted by mother at the age of 2 months. He was admitted for fever, poor urinary stream, and inability to gain weight at the age of 3 months and was evaluated with an ultrasonography (USG) of the abdomen which revealed bilateral hydronephrosis with thickened bladder wall. A micturating cystourethrogram (MCU) revealed small capacity trabeculated bladder with high riding bladder neck

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and dilated posterior urethra suggestive of PUVs. [Figure 1], These were confirmed by cystoscopy and fulguration of the valves was preformed in the same setting, and the patient was discharged in a stable condition. He did well for few months but his symptoms recurred and was admitted at a tertiary care referral center with complaints of fever, urinary dribbling, and failure to thrive. On evaluation the USG showed bilateral hydronephrosis and irregular thickened urinary bladder (wall thickening of 6.5 mm) and bilateral PCN were placed and he was referred to our center with PCN *in situ*. The current haematological evaluation revealed: Hb 7.8 g/dl, total leukocyte count 21,000/mm^3, differential leukocyte count (polymorph 81% and lymphocytes 23%), and platelet count 2.3 lakh/mm^3. Urinalysis revealed protein 2+, pus cells 120–130/Hpf, and red blood cell 8–10/Hpf. Blood urea nitrogen was 11.68 mg/dl while the serum creatinine was 0.2 mg/dl. Urine culture grew *Enterobacter cloacae* which was treated with sensitive parenteral antibiotics. Thyroid function test was normal with a thyroid-stimulating hormone (TSH) level of 2.86 mIU/L. A repeat MCU showed an elongated urinary bladder with trabeculated outline without any vesicoureteric reflux. He also complained of gradually progressive abdominal distension probably due to chronic constipation which was being managed with laxatives and enemas. Barium enema revealed dilated rectum and proximal sigmoid colon with a short-segment persistent narrowing in the region of distal sigmoid colon, suggesting a probability of ultrashort-segment Hirschsprung's disease [Figure 2]. Rectal biopsy confirmed the absence of ganglion cells in the affected segment of the sigmoid colon. Resection of the aganglionic segment followed by anastomosis of normal ganglionic segments was carried out. Postoperative period remained uneventful, and he was started on standard protein energy malnutrition diet protocol with gradual buildup of proteins and calories. Clean intermittent catheterization (CIC) was demonstrated, and the parents were trained to continue it at home. Urodynamic study (UDS) was advised to rule out any bladder storage abnormalities. The parents were advised to follow a strict diet plan with supplementation of essential vitamins and micronutrients. They were further counselled for regular CIC at home and UDS at the next follow-up visit in the pediatric nephrology clinic.

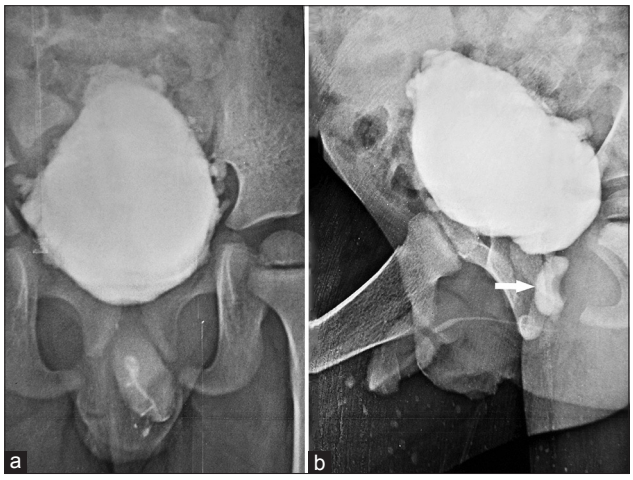

Figure 1: Micturating cystourethrogram showing (a) an elongated hypertrophied bladder with (b) a dilated posterior urethra (arrow)

DISCUSSION

PUVs are detected on around 1 in 1250 screening antenatal USG performed and affect 1 in every 5000 live births.[4] Treatment of PUVs is challenging as it requires active life long management to avoid progressive upper and lower urinary tract deterioration. The few children that survive the initial period also perform poorly with over 50% progressing to ESRD in next 10 years. Therefore, the factors that predispose to further renal damage, such as chronic constipation, must be dealt with seriously. Anatomically, the bladder and the rectum lay in close communication and share the muscular structures of the pelvic floor. Several studies in children have documented the association of constipation with urinary tract abnormalities such as infections, enuresis, vesicoureteral reflux, and upper renal tract dilatation. UDSs have reported the presence of detrusor overactivity in patients with functional constipation.[5] This coexistence of bladder and bowel dysfunction may have several proposed etiologies: It may be that (a) rectum and bladder are affected by the same neuropathology; (b) dysfunction in one system may impede the neighbouring organ structures mechanically, e.g., stool impaction from severe constipation may impede voiding; or (c) severe straining due to constipation may induce changes in the pelvic floor musculature and cause urinary incontinence.[2] In children with constipation with concomitant LUTS, adequate treatment of constipation also improves or abolishes LUTS. Hirschsprung's disease is a rare disease, which affects 1 in 5000 live births and is a consequence of premature arrest of craniocaudal migration of neural crest-derived neuroblasts in the hindgut.[6] Careful history including delayed passage of meconium at birth and subsequent bowel patterns has been quite helpful in it's

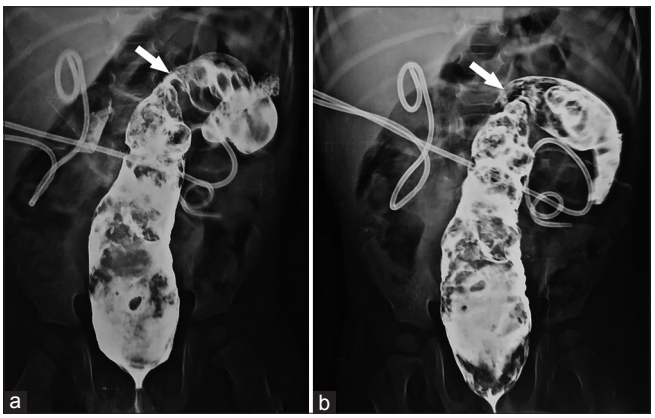

Figure 2: Barium enema (a and b) showing dilated rectum and proximal sigmoid colon with a short segment focal area of narrowing (arrows) in the region of sigmoid colon with bilateral percutaneous nephrostomy tubes *in situ*

early diagnosis and timely management. Only a handful of studies are available in the literature describing the association of Hirschsprung`s disease with CAKUT. Pini Prato *et al*. found a 20.7% incidence of CAKUT in their cohort of patients with Hirschsprung's disease, but only one patient in this cohort had PUV.[3] As the incidence of both PUV and Hirschsprung's disease is quite rare in isolation, their combination is an extremely rare entity.

It is important to elicit detailed history and perform focussed clinical examination to asses bowel habits in children with LUTS, as constipation may not be recognised and thus may not be reported by the parents. Early cystoscopic ablation of PUV and timely surgical intervention for Hirschsprung's disease would definitely delay the progression of ESRD in such an unusual and entirely unwanted association.

CONCLUSION

Sometimes, questioning the parents about the bowel habits of the child is not enough as constipation is frequently not recognized and reported by the parents. It is important to pay attention during the physical examination to the findings suggestive of stool impaction, such as abdominal masses and/or abdominal distension. In a child presenting with features of LUTO, causes for constipation must be looked for on a regular basis. Ignoring pathological constipation, which may significantly contribute to progression of CKD, can prove hazardous.

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