Obstruction of the ureter by neurofibroma of the urinary bladder treated endoscopically

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ABSTRACT

We present a case of a 23-year-old male with NF-1 diagnosed with bladder neurofibroma at childhood with regular ultrasound (US) follow-up since then, who presented with an obstructed left kidney. A detailed evaluation including blood tests and advanced imaging revealed left hydroureteronephrosis associated with a large bladder mass. The patient underwent cystoscopy and resection of the left ureteral orifice that was covered by the mass, and an indwelling ureteral stent was left in place. A follow-up US was performed one month after stent removal showing resolution of the hydronephrosis.

1. Introduction

Neurofibromatosis type 1(NF-1) is an autosomal dominant transmitted disease with various clinical presentations including skin pigmentation, hamartomas located within the iris (Lisch nodules) and multiple benign neurofibromas. Other manifestations include learning disabilities, vascular disease, benign central nervous system tumors and skeletal anomalies.

Genitourinary system involvement is extremely rare, and less than 80 cases were reported in the literature with the bladder being the most common affected organ. Other organs that may be involved include the prostate, the penis, the spermatic cord, the urethra, the ureters, the testicles and the clitoris.

When the bladder is affected, the most common manifestations are recurrent urinary tract infections, irritative symptoms, hematuria and urinary tract obstruction.

We present a case of a bladder neurofibroma in a patient with NF-1 associated with obstruction of the left ureter treated endoscopically.

2. Case presentation

A 23-year-old man with a history of NF-1 and a known bladder neurofibroma since childhood was referred to our clinic due to a new onset left hydronephrosis. The bladder neurofibroma was initially diagnosed at childhood and was followed on a yearly basis by ultrasonography (US) ever since and did not show any growth pattern throughout the years. His other NF-1 manifestations were multiple café-au-lait macules, Lisch nodules, optic glioma of the left optic nerve and cutaneous neurofibromas.

An US performed several days prior to his referral (Fig. 1) revealed dilation of the left renal pelvis and ureter up to the distal part and a growing bladder mass. He had no urinary tract symptoms nor hematuria and his renal function was preserved on blood tests.

The patient completed a computed tomography urogram that revealed left hydroureteronephrosis associated with a large bladder mass (Fig. 2). He subsequently underwent cystoscopy that showed a 2–3 cm width mass infiltrating the posterior bladder wall extending from the dome to the bladder trigone (Fig. 3). The left ureteric orifice was identified only after an intravenous injection of Indigo Carmine. Resection of the orifice was performed and exposed a patent left ureter. A ureteral stent was left indwelling and removed after one week. A follow-up US one month later showed complete resolution of the hydronephrosis. The specimen demonstrated pathologic changes diagnostic for neurofibroma.

3. Discussion

NF-1 incidence is 1 in 3000 live births. It is caused by a dominant loss of function mutations in the NF-1 gene located on the long arm of chromosome 17. However, more than half the cases are sporadic,
representing de novo mutations. The male to female ratio is 3:1.

The disease is clinically diagnosed when any 2 of the following 7 features are present: (1) six or more café-au-lait macules larger than 5 mm in greatest diameter in prepubertal individuals and larger than 15 mm in greatest diameter in postpubertal individuals. (2) Axillary or inguinal freckling. (3) Two or more iris Lisch nodules (hamartomas). (4) Two or more neurofibromas or 1 plexiform neurofibroma. (5) distinctive osseous lesion such as sphenoid dysplasia or cortical thinning of long bones with or without pseudoarthrosis. (6) Optic gliomas. (7) A 1st-degree relative with NF-1 whose diagnosis was based on the aforementioned criteria.

Malignant transformation of neurofibromas is estimated to occur in approximately 3–5% of patients, and it should be suspected when there is progressive enlargement or pain associated to the mass. In addition, irregular infiltrative borders and internal inhomogeneity at imaging should raise the possibility of malignant transformation.

Neurofibromas of the genitourinary tract are extremely rare and less than 80 cases were described in the literature. They can involve the prostate, urethra, testis, spermatic cord, clitoris and the ureter, but the bladder is the most common site. These tumors commonly arise from the pelvic, bladder nerves and prostatic nerve plexus. In the bladder, they arise from the nervous ganglia of the bladder wall. Common presenting clinical features include recurrent urinary tract infections, hematuria, irritative symptoms and urinary tract obstruction. The patient discussed in this case had a unilateral urinary obstruction secondary to the bladder neurofibroma, but he was completely asymptomatic.

Bladder neurofibromas are usually treated conservatively unless
there is urinary tract obstruction, refractory urinary symptoms or hematuria, significantly decreased bladder capacity or if there is suspicion for malignant transformation of the tumor. There is no standard of treatment and it depends on the extent of disease and the reason for intervention. Treatment options include transurethral resection (TUR), partial cystectomy and radical cystoprostatectomy with urinary diversion. In our case, a TUR of the left ureteric orifice was performed due to its obstruction by the mass. On a follow-up US the hydronephrosis was resolved. However longer followup is warranted in order to decide if a more radical treatment is needed.

4. Conclusion

Bladder neurofibromas are usually benign lesions with good prognosis, and when asymptomatic surveillance is the treatment of choice. In our patient, a large neurofibroma obstructed the left ureteric orifice and caused hydroureteronephrosis, thus treatment was warranted. We chose to treat him endoscopically with TUR and on a follow-up US the hydronephrosis was resolved. Longer followup is needed.

Consent

This case report has no identifying details nor identifying figures of the patient, thus consent was not warranted.

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Declaration of competing interest

No conflict of interest.

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