An isolated case of an incidentally discovered neurofibroma of the urinary bladder

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ABSTRACT

Neurofibromas of the bladder are extremely rare tumors formed in peripheral nerves and are generally considered to occur in-conjunction with neurofibromatosis type 1 (NF-1). The following is an isolated case of a 19-year-old, asymptomatic male patient presenting with bladder tumor discovered incidentally on CT-scan. The patient had no known family history or any exam findings consistent with NF-1. The patient underwent transurethral resection of the tumor and pathologic analysis confirmed the final diagnosis of neurofibroma of the bladder. The patient’s course, procedure and treatment are reported.

Introduction

Neurofibroma of the bladder was originally described by Gerhardt during an autopsy in 1878. The bladder is the most commonly affected organ in the urinary tract with around 70 cases reported in the literature. Neurofibromas of the genito-urinary tract have been shown to derive from nerves of the pelvic, vesical and prostatic plexuses. In the bladder, neurofibromas arise in the nervous ganglia of the bladder wall.

Neurofibromas and schwannomas can be more broadly categorized as epithelioid sheath tumors. These tumors are the most common forms of nerve sheath tumors and are generally associated with Neurofibromatosis (NF). Isolated cases in the urinary bladder are very rare and contribute to less than 0.1% of all bladder tumors.

Neurofibromas are generally diagnosed through physical examination, and cross sectional imaging. If the neurofibroma is focal, conservative excision is the choice of treatment with a low risk of recurrence, however diffuse neurofibromas may require more extensive excision. Diagnosis can be confirmed histologically as bladder involvement is usually generalized with diffuse proliferation of uniform neurofibroma cells. Neurofibromas almost always occur in the setting of Neurofibromatosis type 1 (NF-1), therefore any sporadic cases of neurofibroma justifies genetic screening.

Case description

We report a 19-year-old male who was admitted after a physical altercation. He underwent CT imaging of the abdomen/pelvis that incidentally revealed a 2.4cm mass with apparent right seminal vesical involvement and encroachment upon his urinary bladder (Fig. 1). Patient had a follow-up MRI which showed an intravesical mass (Fig. 2). He was subsequently referred to our urology center. During urologic consultation, the patient denied any lower urinary tract symptoms such as hematuria, dysuria, or urinary retention. Additionally, the patient denied any family history of NF-1 and showed no clinical manifestations of NF-1 such as cafe-au-lait pigmentation, or Lisch nodules.

Patient presented for diagnostic cystoscopy discovering a mass on the right lateral trigone that appeared to be pushing into the bladder. The ureteral orifices were uninvolved. A trans-rectal ultrasound clearly identified the mass, and it was elected to have the tumor resected. Tissue was sent for histopathologic analysis which showed benign spindle cell neoplasm, positive for S-100, CD34 and beta-catenin, with mast cells (Fig. 3). The pathologic report was most suggestive of neurofibroma of the bladder.

At follow up, the patient noted that he had tolerated the procedure well and denied any hematuria or urinary retention. He noted a small period of dysuria which was self-limiting and resolved. The patient denied any family history of NF-1 but does report a cousin diagnosed with renal cell carcinoma having undergone a nephrectomy. The patient has no other known genetic syndromes in the family. It was recommended that the patient undergo genetic screening based on the findings of neurofibroma.

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Discussion

Isolated neurofibroma of the bladder is very rare and account for less than 0.1% of all bladder tumors. This disease is generally found in conjunction with the genetic disease NF-1, and urinary neurofibromas are 3-fold more common in males than females. Since this is a rare disease, it may be difficult to diagnose. A differential diagnosis of leiomyosarcoma, ganglioneuroma and paraganglioma should always be
Neurofibroma of the bladder generally presents in childhood or young adulthood. In a study of neurofibroma of the bladder at Mayo Clinic from 1965 to 1990, they reported four cases of neurofibroma of the bladder with a mean age of 17 at diagnosis. The patient discussed here was diagnosed at age 19, which correlates with previous observations of the disease. Additionally, it was noted that none of the cases in the four studied resulted in malignant transformation after a ten year follow-up period. Studies involving neurofibroma of the bladder report malignant transformation in 5–10% of patients, while others studies of non-genitourinary neurofibromatosis note that malignant transformation in 12–29% of patients.

Some of the most common symptoms of urinary involvement of neurofibroma are hematuria, dysuria, and irritative complaints with varying severity. Some cases may also note dysfunctional voiding or urinary retention related to location of the mass near the bladder neck. The patient discussed in this case had a bladder mass that was discovered incidentally, and was asymptomatic. Neurofibroma will present itself differently in each patient varying on the size and number of tumors, but generally symptoms are directly related to mass effect.

The patient’s neurofibroma was managed surgically in order to obtain a pathologic diagnosis. Neurofibroma was confirmed histologically following TURBT. Normally, a patient with a single neurofibroma experiencing few urinary symptoms would be dealt with more conservatively.

Although neurofibromas are most generally found in conjunction with NF-1, sporadic cases of neurofibroma have been reported in patients with no other stigmata of the disease. The patient discussed here denied any family history of NF-1 and showed no signs of the genetic disease, thus it is highly recommended that the patient proceed with genetic screening for NF-1. There are no follow up guidelines specific to this disease, however we recommend the patient’s follow up should include routine imaging to monitor for local recurrence given the chance for malignant transformation.

**Conclusion**

We present a 19 year-old male with an incidentally discovered bladder mass that was diagnosed as an isolated neurofibroma. While our patient had no NF-1 diagnosis, his presentation raises some suspicion. Isolated neurofibromas of the bladder are extremely rare, therefore management recommendations are largely extrapolated from other

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**Fig. 3.** A. Low magnification, H&E stain: Bladder tumor (left/a) and smooth muscle of bladder wall (right/b). B. Desmin stain: bladder wall. C. Low magnification, S-100 stain: tumor. D. High magnification, H&E stain: tumor. E. High magnification, S-100 stain: tumor.
bladder tumors. Transurethral resection of the tumor is appropriate to establish diagnosis in a patient without symptoms. If the patient is symptomatic or has widespread disease, a more complete resection of the tumor is recommended. Close follow-up with the patient in the clinic is essential and lower urinary tract symptoms should continue to be monitored, as urinary tract symptoms may be the initial indication of recurrence. Follow up cystoscopic evaluation should be coupled with imaging studies to monitor for potential local recurrence or progression.

Declaration of competing interest

The authors declare no conflict of interest.

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