Bladder Neurofibromatosis Causing Urinary Tract Obstruction: Case Report

Üriner Sistem Obstrüksiyonuna Neden Olan Mesane Nörofibromatozisi

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ABSTRACT Neurofibromatosis is an autosomal dominant disease characterized by developmental abnormalities of skin along with an increased risk of nervous system tumors that may become cancerous. Urinary system involvement in neurofibromatosis is a rare condition. Genitourinary tract neurofibromas usually arise from the pelvic and bladder nerves, and the prostatic plexus. Bladder is the most affected organ in the urinary tract. Bladder neurofibromatosis may present as a diffuse infiltrative process or an isolated neurofibroma. Diffuse neurofibromas can be difficult to diagnose leading to delayed treatment and potential need for more extensive excision. We report our experience and management of a case with bladder neurofibromatosis with upper urinary tract and bladder outlet obstruction.

Key Words: Neurofibromatosis 1; urinary bladder neoplasms; ureteral obstruction; urethral obstruction; therapy

ÖZET Nörofibromatozis, derinin gelişimsel anomalileri, kansere dönüşebilen sinir sistemi tümörleri riski ile karakterize otozomal dominant bir hastalıktır. Nörofibromatoziste üriner sistem tutulumu nadir görülen bir durumdur. Genitoüriner sistem nörofibromları genellikle pelvik, mesane sinirlerinden ve prostatik pleksustan kaynaklanır. Üriner sistemde mesane en çok etkilenen organdır. Mesane nörofibromatozisi diffüz infiltratif veya izole nörofibrom olarak ortaya çıkabilir. Diffüz nörofibromlarda tanı koymadaki zorluk tedavinin gecikmesine ve daha geniş eksizyon gereksinimine neden olabilmektedir. Bu olguda, üst üriner sistem ve mesane çıkım obstrüksiyonuna neden olan bir mesane nörofibromatozis tecrübemiz ve yaklaşımımız sunulmuştur.

Anahtar Kelimeler: Nörofibromatozis 1; mesane tümörleri; üreter tıkanıklığı; üretra tıkanıklığı; tedavi

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Two types of neurofibromatosis have been defined. The type I or Von Recklinghausen's disease is characterized by benign peripheral nerve tumors called neurofibromas and pigmented skin lesions called 'cafe au lait spots' located at non-sunexposed areas like axilla are the predominating clinical manifestations. Other clinical findings are hamartomas of the iris called 'Lisch nodules' and pseudoarthrosis of the tibia. Compression to the nervous system may lead to radiculopathy, neuropathy and aqueductal stenosis may cause hydrocephalus, scoliosis, short stature, hypertension, epilepsy, and mental retardation. Patients with type 1 neurofibromatosis have increased risk of developing nervous system tumors

like plexiform neurofibromas, optic gliomas, ependymomas, menengiomas, astrocytomas and pheochromocytomas.[1] Type 2 neurofibromatosis is mainly characterized by the development of bilateral vestibular schwannomas.[2] There is increased risk for the development of meningiomas, gliomas and schwannomas of cranial and spinal nerves.

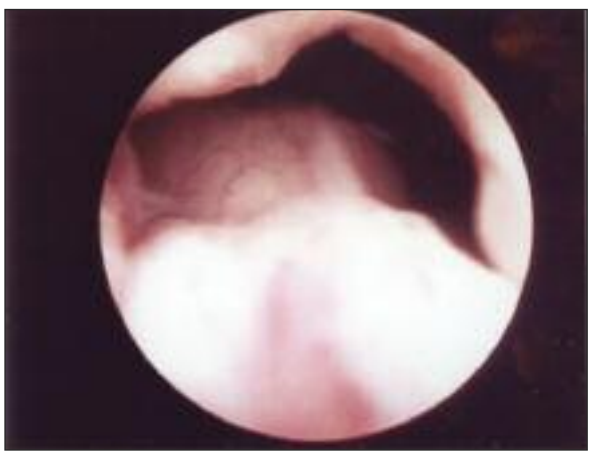
Neurofibromatosis rarely involves the upper urinary tract and the bladder.

CASE REPORT

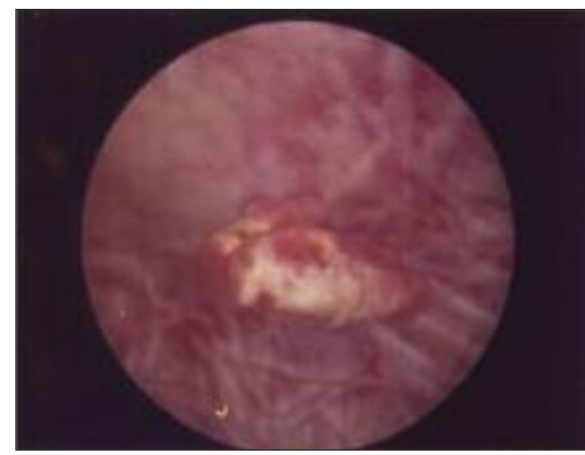
A 33-year-old female was admitted to our clinic with complaints of frequency, urgency, low urinary stream and lumbar pain. Her medical history revealed diagnosis of a bladder mass at age 7, but the family had rejected any intervention. She was operated for vascular stenosis of a cranial vessel at age 24. On physical examination, she was pale and had cafe au lait spots all over the body. Blood pressure was 80/40 mmHg. There was a low grade systolic murmur at mitral area. The suprapubic area was tender on palpation. Lisch nodules were seen on eye examination. Physical examination of the other organ systems revealed no abnormality. Routine blood chemistry were within normal limits but hemoglobin was found to be 8.85 g/dL. Further diagnostic work-up for anemia revealed iron and vitamin B12 defficiency. Abdominal ultrasound demonstrated mild hydroureteronephrosis on the left side and significant dilatation in the right kidney that was managed with percutaneous nephrostomy. Abdominal computed tomography revealed bladder wall thickening, a 4 x 2 cm irregular solid mass inferior to the bladder at the level of distal ureters and a 2.5 x 1 cm polypoid mass on the posterior wall of the bladder. Uroflowmetric parameters with a voided volume of 196 mL were 12 mL/sec peak flow rate; 7 mL/sec average flow rate and 54 mL residual urine. Repeat uroflowmetry revealed similar findings. A mass on the trigone bulging into the lumen, involving the ureteral orifices and another polypoid mass on the posterior bladder wall were seen on cystoscopic examination (Figure 1a, 1b). Following resection of the masses the left ureteral orifice became visible. Lich-Gregoir ureteroneocystostomy was performed to relieve the right hydroureteronephrosis after two weeks. Histopathological examination revealed neurofibroma (Figure 2a-d). There was no malignant transformation. On the postoperative sixth week intravenous urography showed normal left kidney and moderate hydroureteronephrosis on the right side. MAG-3 renal scans revealed non-obstructive dilatation on the right side.

DISCUSSION

Manifestations of neurofibromatosis in the skin, the eye, the skeletal and nervous systems have been well documented since the disease was first described by Smith in 1882. Urological manifestations of neurofibromatosis are rare. Up to date, the most commonly reported site of presentation is the blad-

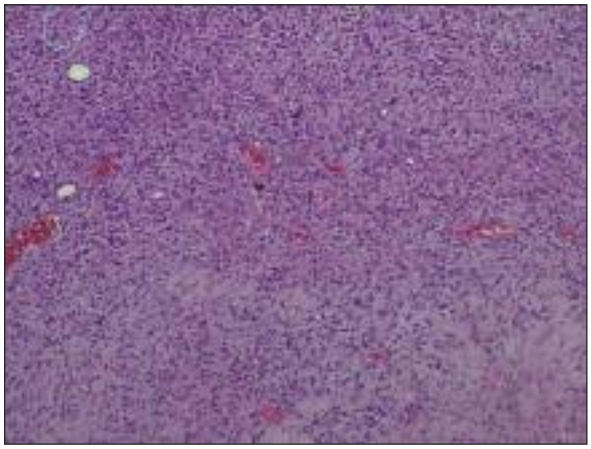


a

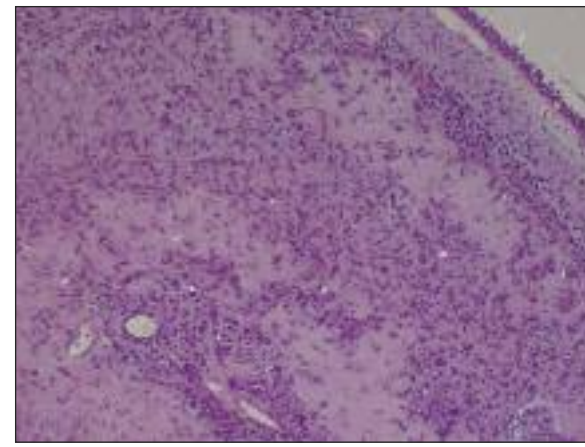


b

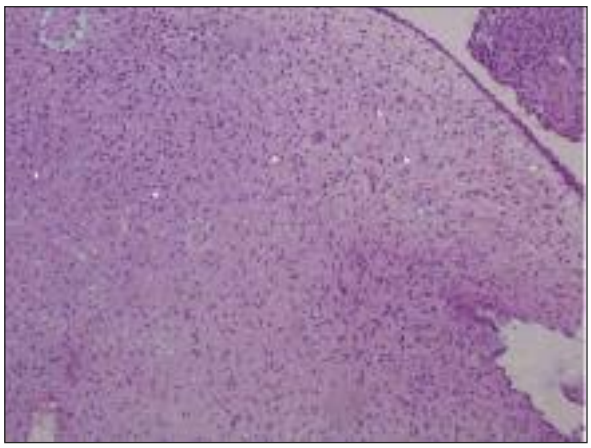
FIGURE 1: Endoscopic view of the mass on the trigone bulging into the lumen **(a)** and the polypoid mass on the posterior bladder wall **(b)**.



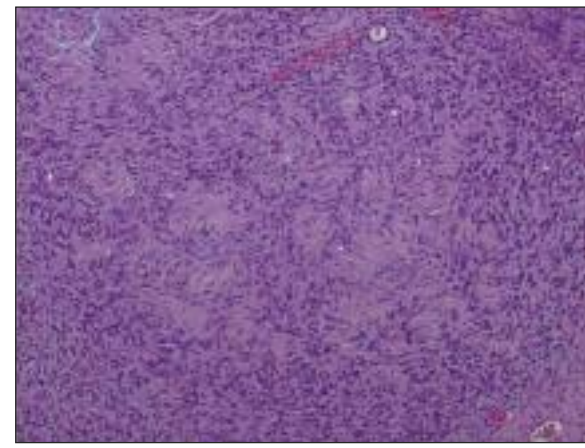
a: (Haematoxylin and eosin, x10)



b: (Haematoxylin and eosin, x10)



c: (Haematoxylin and eosin, x10)



d: (Haematoxylin and eosin, x20)

FIGURE 2: A tumoral mass composed of fascicles of elongated spindle-shaped cells with thin wavy nuclei in collagenized and fibrillar background in the bladder **(a, b)**. Within the mass, myxoid areas were also present **(c)**. Wagner-Meissner bodies were seen **(d)**.

der, that was first reported by Gerhardt in 1878.[3-6] Bladder neurofibromas originate from the nerve sheaths of the autonomic nerve plexus of the bladder, bladder neck and prostate. Incidence of malignant degeneration has been reported as 12 to 29% and only four cases of malignant bladder neurofibromas exist in the literature.[5-7] The most common symptoms of bladder neurofibroma are irritative and obstructive complaints. Hematuria, incontinence, enuresis, acute urinary retention, flank pain, palpable mass are the other reasons for admitting to urology outpatient clinics. Bladder neurofibromas may present as a solitary mass within the bladder wall or as a diffuse infiltrative form. Upper urinary tract obstruction is generally associated with extensive disease. Close follow up is necessary to detect upper tract obstruction. Although conservative approach is proper for localized lesions without upper tract obstruction, transurethral resection or partial cystectomy is the suggested treatment modality by some authors. Permanent urinary diversion may be indicated to prevent renal deterioration in patients with severe upper tract obstruction and who have been treated with temporary urinary diversion or ureteral reimplantation.[8] Malignant transformation should be considered in patients with progressive tumor growth.

Urinary tract obstruction may be a manifestation of neurofibromatosis. Complete resection of neurofibromas may not always be possible but relief of obstruction could be achieved.

As has been observed in the presented case urinary tract involvement in neurofibromatosis can display a long course before causing significant obstruction and symptoms.

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