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Case Report

Neurofibromatosis Type 1 with Prostate Involvement Presenting As Urinary Retention - A Rare Case Report

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ABSTRACT

Neurofibromatosis type 1 (NF1) is an autosomal dominant disease with various clinical manifestations. Neurofibromas can involve genitourinary tract such as penis, clitoris, prostate, urethra, testes, spermatic cord, ureter but most commonly affects urinary bladder. NF1 is more common in males with ratio of 3:1. Malignant transformation in generalized NF is rare accounting 12% - 29%. The treatment option for benign disease is usually conservative. The size and location of the mass determine the symptoms. We present a 23 year old male patient with NF1 involving prostate and resulting in urinary retention. This is rare and yet to be published in the literature.

Key words: Neurofibroma, Prostate enlargement, urinary retention

INTRODUCTION

NF1 is commonly associated with peripheral nerve sheath tumors, whereas NF2 primarily affects the central nervous system. [1] Alterations of skin pigmentation, iris Lisch nodules, and multiple benign neurofibromas usually constitute the clinical picture. However, patients also have learning disabilities and may develop skeletal abnormalities, vascular disease, central nervous system (CNS) tumors, or malignant peripheral nerve sheath tumors. [2]

Genitourinary tract is rarely involved in NF1 and less than 80 cases were reported in the literature to date. [3] The bladder is the commonest affected organ in the urinary tract. [4] Approximately less than one-third of these cases are in pediatric population. The most frequent symptoms of these patients are recurrent urinary tract infection, hematuria, and irritative symptoms. Urinary retention and constipation due to the mass compression on intestinal structures are relatively rare findings in NF1 with genitourinary tract involvement. [5] Neurofibromas can involve genitourinary tract such as penis, clitoris, prostate, urethra, testis, spermatic cord, and ureter. However, NF most commonly affects the bladder. [6] The condition is more common in males than female by a ratio of 3: 1. Mutations of the NF1 gene which is located on chromosome 17 lead to abnormal tumor suppression. Consequently, patients with NF1 have an increased prevalence of benign and malignant neoplasms. We aim to present

an adolescent patient with prostate involvement presenting as dribbling and urinary retention.

CASE HISTORY

A 23 year male presented to our hospital with history of dysuria, frequency, thin stream, incomplete emptying of bladder, and change in gait (limping) since 6 months, dribbling of urine since 1 month and retention since 1 day. We catheterized the patient. He had undergone right herniorrhaphy 2 yrs back. His medical history was unremarkable.

On examination, multiple café au lait spots were present on his body (Fig 1), with swelling in the posterior aspect of both the thighs and medial aspect of both arms (Fig 2, 3) palpation of which caused tingling and pain. B/L proptosis was present. Multiple swellings could be palpated in the posterior triangle of neck and abdomen. Power in the upper limb and lower limb muscles were normal. Per rectal examination revealed grade 2 prostatomegaly with normal sphincter tone. External genitalia examination was normal.

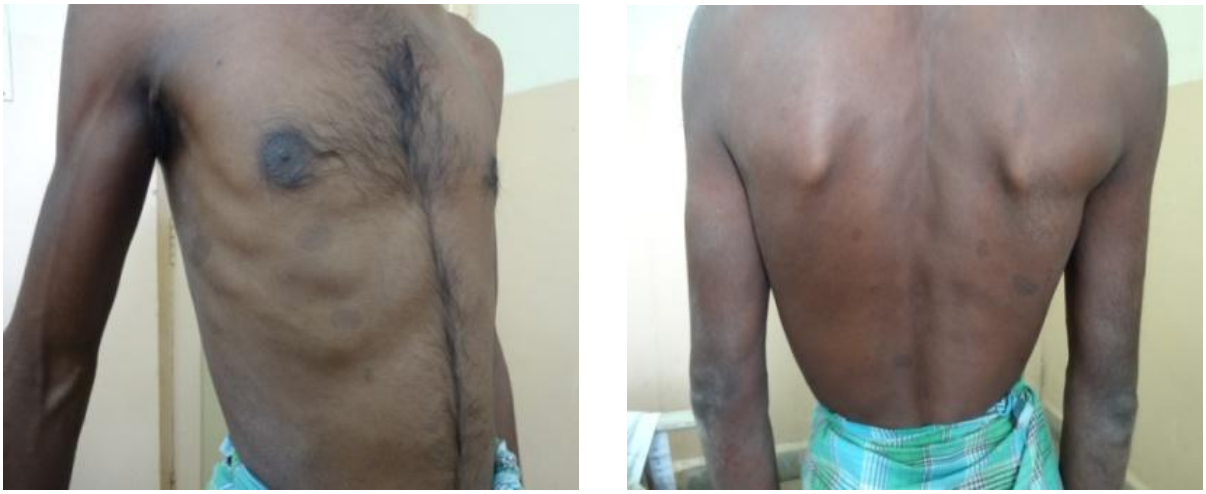

Fig 1: Cafe-au- lait macules over lumbar area

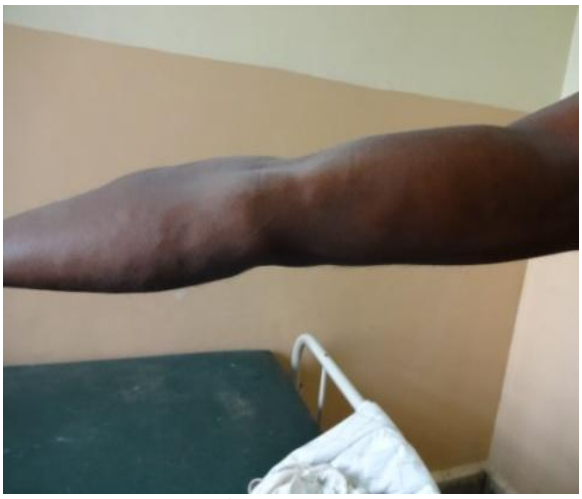

Fig 2: Neurofibromas along ulnar nerve

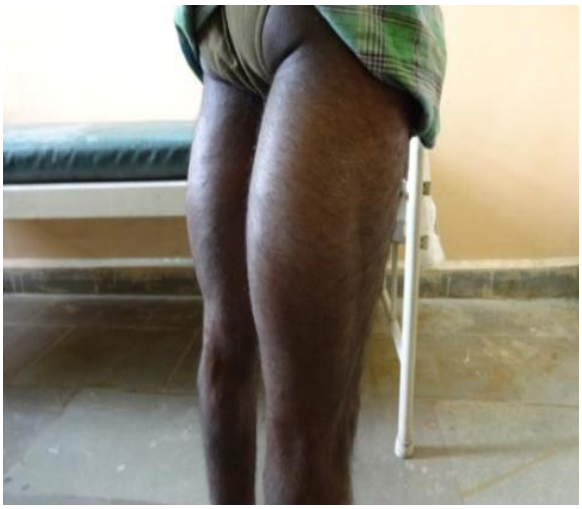

Fig 3: Bilateral thickened sciatic nerves

Fundus examination was normal. Ultrasound revealed normal kidneys & bladder with multiple enlarged soft tissue masses in the abdomen and pelvis resembling lymph nodes, enlarged prostate with heterogenous echotexture weighing 60 gms with pre-void residue of 150 cc. X-ray chest and skull, CT brain were normal. FNAC of the neck swelling revealed haemorrhagic aspirate. Contrast CT scan and MRI of abdomen, pelvis, limbs (Fig 4, 5, 6) showed plexiform neurofibromatosis.

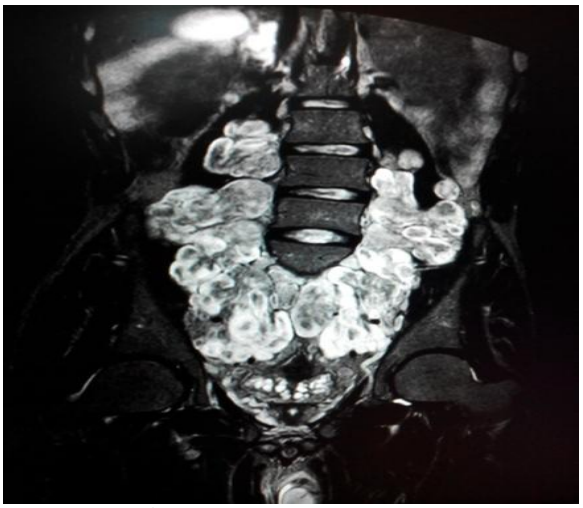

Fig 4: MRI of the abdomen & pelvis showing extensive plexiform neurofibromas involving bilateral paraspinal nerves, lumbar plexes on STIR coronal images

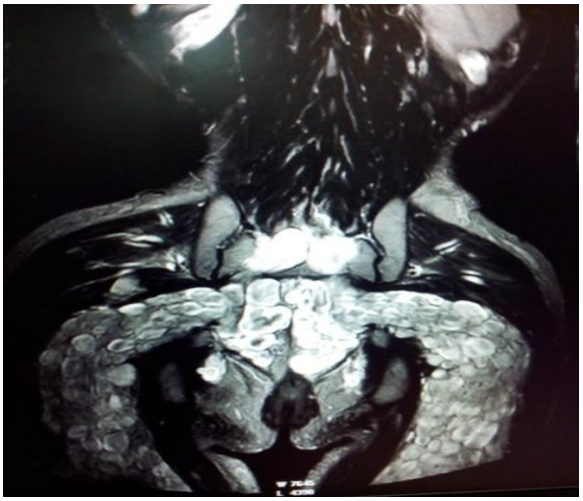

Fig 5: MRI of the pelvis showing extensive plexiform neurofibromas involving bilateral sciatic nerves on STIR coronal images

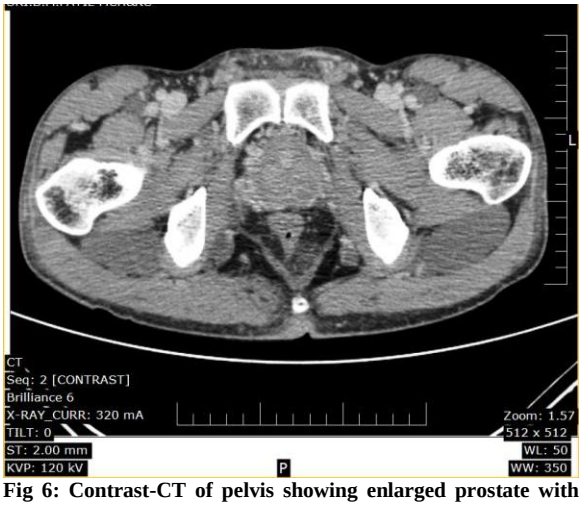

Fig 6: Contrast-CT of pelvis showing enlarged prostate with heterogenous enhancing areas

DISCUSSION

NF1 is one of the most common genetic disorders affecting approximately 1 in 3000 patients. Neurofibromatosis 1 (NF1) is an autosomal dominant disease caused by heterozygous mutations of the NF1 gene, which is located on chromosome 17, lead to abnormal tumor suppression. Consequently, patients with NF1 have an increased prevalence of benign and malignant neoplasms. Neurofibromas are benign nerve sheath tumors and the hallmark lesion of the NF1. Plexiform neurofibromas are pathognomonic for NF1, usually involving a long segment of a major nerve trunk and extending into the nerve branches and they result in the so-called bag of worms appearance on gross inspection and cross-sectional imaging. NF1 is commonly associated with peripheral nerve sheath tumors, whereas NF2 primarily affects the central nervous system. Most patients with NF1 have only milder manifestations of the disease, such as pigmentary lesions and Lisch nodules.

Genitourinary tract is rarely involved in NF1 and less than 80 cases are reported in the literature to date. The bladder is the commonest affected organ in the urinary tract but can involve penis, clitoris, prostate,

urethra, testis, spermatic cord, and ureter. Approximately less than one-third of these cases are in pediatric population. The most frequent symptoms of these patients are recurrent urinary tract infection, hematuria, and irritative symptoms. Urinary retention and constipation due to the mass compression on intestinal structures are relatively rare findings in NF1 with genitourinary tract involvement. [7] Only five cases of solitary renal parapelvic or parenchymal neurofibromas have been reported in the literature. [8-12] Krishna et al. reported a patient with neurofibromatosis suffering urinary retention as an initial symptom. [13]

The diagnosis of NF1 is largely based on clinical criteria established by the National Institutes of Health Consensus Development Conference, [14] that is, the presence of two or more of the following: cafe-au-lait macules or neurofibromas, Lisch nodules, axillary or inguinal freckling, optic glioma, distinctive osseous lesions or first-degree relatives with NF1. However, patients also have learning disabilities and may develop skeletal abnormalities, vascular disease, central nervous system (CNS) tumors, or malignant peripheral nerve sheath tumors. Because these clinical criteria are well established and widely accepted, pathological confirmation of neurofibroma is not a requirement and is not routinely recommended for the diagnosis of NF1. Our patient had 2 criteria for NF1 i.e., Café-au-lait macules (more than 12) and neurofibromas

A systemic physical examination may facilitate the diagnosis of NF if cafe-au lait spots and superficial neurofibromas are present and recognized. Imaging techniques are useful for assessing the manifestations of neurofibromatosis especially for the lesions in abdominopelvic and cranial regions.

Computed tomography (CT) of plexiform neurofibromas shows large multilobulated low-attenuation masses usually within a major nerve distribution. The attenuation values range from 20 to 25 HU on non-enhanced scans and 30–50HU on intravenous contrast-enhanced scans. [15] The low attenuation of neurofibromas has been attributed to myxoid and mucinous stroma that can be observed microscopically within these tumors. [16] Tonsgard et al reported intravenous contrast enhancement in 50% of their patients with abdominal or pelvic plexiform neurofibromas. [17] This enhancement may be homogenous or heterogenous. MRI reveals large conglomerate masses consisting of innumerable neurofibromas, diffusely thickening the involved nerve and often extending into nerve branches. The MRI features of neurofibromas are characteristic and can be helpful in the evaluation of a mass in a patient with known NF1. Neurofibromas show characteristically low signal intensity on T1 weighted images and heterogenous high signal intensity on T2 weighted images. The high T2 signal corresponds pathologically to areas of cystic degeneration or myxoid matrix and the low T2 signal represents collagen and fibrous tissue. [18] The areas of low T2 signal enhance following gadolinium administration. Plexiform neurofibromas have a characteristic ringlike or septated pattern that represents the complex fascicular arrangement typical of these tumors. [19] The rate of malignant transformation of neurofibromas has been estimated at 12–29% in patients with neurofibromatosis-1. [20]

Our patient had a large asymmetric and heterogeneous mass in CT and MRI scans of the abdomen, neck. Prostate showed enlargement with heterogenous density in CT. In our patient, we placed him on α-blocker & trial voiding was successful. On follow-up at 3 months, he is doing well and is asymptomatic. The prognosis of

neurofibromas is generally very good with a very rare malignant transformation rate. Neurofibroma with prostate involvement and urinary retention is rare & yet to be published in literature. Hence we are reporting this case.

CONCLUSION

Prostate involvement in neurofibromatosis is extremely rare. A young patient presenting with urinary retention should also remind you the rarest possibility of NF prostate. Only one case of neurofibromatosis with urinary retention has been mentioned in the literature but there are no reports of neurofibromatosis with prostate involvement leading to urinary retention being mentioned.

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