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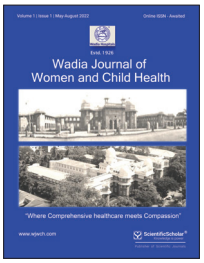

Case Report

Pediatric genitourinary plexiform neurofibroma in a case of neurofibromatosis type 1

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Received: 26 May 2024
Accepted: 21 December 2024
Published: 15 March 2025

DOI
10.25259/WJWCH_28_2024

ABSTRACT

Genitourinary plexiform neurofibromas are rare presentation in patients with neurofibromatosis 1. We present a rare case of a 10-year-old girl who presented with abdominal pain and menorrhagia due to a large infiltrative plexiform neurofibroma with completely replaced uterine myometrium with involvement of other pelvic organs.

Keywords: Pediatric genitourinary plexiform neurofibroma, Genito-urinary system, Neurofibromatosis type 1

INTRODUCTION

Von Recklinghausen disease, commonly known as neurofibromatosis type 1 (NF1) is an autosomal dominant disease that shows incomplete penetrance related to a mutation in the neurofibromin gene on chromosome 17q. The presence of cutaneous symptoms is a benign finding whereas visceral involvement in the form of plexiform neurofibromas is rare with a dubious and progressive course and presents a great challenge for management.[1] Patients usually present with urinary infections and males are affected more than females.[2] Genitourinary involvement is infrequent with an incidence of 2 per 361 patients[3] and most commonly involves the urinary bladder[4,5] followed by urinary tract and then genitalia.[6,7] Neurofibromas of the female genital tract are extremely uncommon. Vulva is the most frequent genital organ affected.[8,9]

The diagnosis is established on histology, but the extent and nature of lesions can be reliably assessed on MRI due to its higher soft-tissue resolution. The classically described "target sign" is seen as a low signal center surrounded by hyperintense signal. In addition, MRI can be used for surveillance and growth monitoring. Computed tomography is used when osseous involvement is suspected.

The management is mostly conservative with surgery offered only in cases presenting with severe mass effect, although complete resection is rarely achieved due to the infiltrative nature of the plexiform neurofibroma. Periodic clinical and radiological monitoring should be performed for early diagnosis and detection of early features of malignant transformation.[10]

How to cite this article: Pathak P, Gala F. Pediatric genitourinary plexiform neurofibroma in a case of neurofibromatosis type 1. Wadia J Women Child Health. 2024;3:137-40. doi: 10.25259/WJWCH_28_2024

CASE REPORT

A 10-year-old girl presented with abdominal pain for 1 year which was dull aching, diffuse, and associated with menorrhagia and occasional spotting. There were no urinary or bowel-related complaints according to the patient. The child was adopted at 1 year of age, and hence, detailed birth/family history was not known; however, developmental milestones were achieved age appropriately. On physical examination, multiple hyperpigmented café-au-lait spots [Figure 1] were seen all over the body, ~ 15 mm in size with presence of axillary freckling. Abdominal examination revealed periumbilical tenderness with no soft-tissue swelling. On ultrasonography (USG), a large infiltrative heteroechoic mass was seen infiltrating the uterus replacing the myometrium completely and abutting a thin stripe of endometrium [Figure 2]. The mass was seen infiltrating the posterior bladder wall. Magnetic resonance imaging (MRI) confirmed these findings of diffuse enlargement of the uterus due to large infiltrative T1 hypointense/T2 hyperintense/heterogeneously enhancing plexiform neurofibroma which also involved the cervix and vagina, infiltrated the posterior wall of the urinary bladder, and encased the urethra with classical target appearance [Figure 3]. It also encased the rectum and involved the perirectal fat, and presacral fat; extending inferiorly into vulva with enlargement of clitoris [Figure 4]. Bilateral ovaries were not involved.

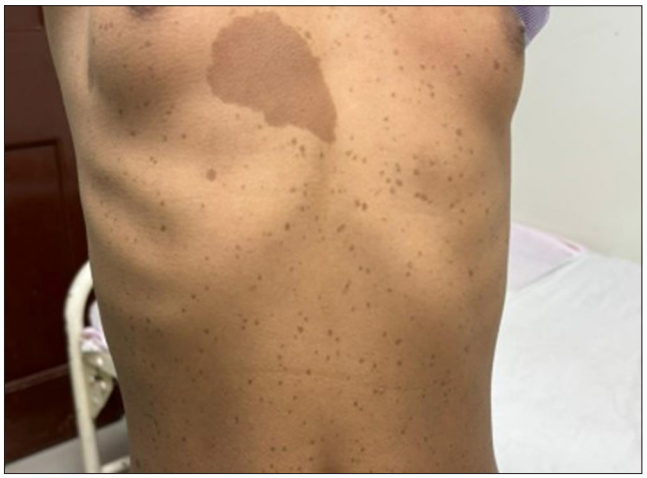

Figure 1: On examination, multiple café-au-lait spots were seen all over the body, approximately more than 15 mm in size.

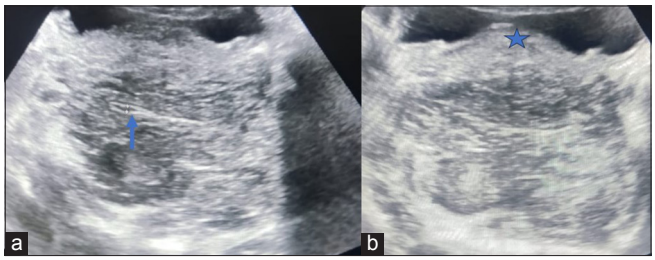

Figure 2: (a-b) Ultrasound images depicting complete replacement of myometrium by hetero-echoic soft tissue abutting endometrium (blue arrow) and invading posterior bladder wall (blue star).

Imaging of the brain revealed multiple ill-defined T2/fluid-attenuated inversion recovery hyperintense foci in bilateral globi pallidi, thalami, midbrain, pons, right half of medulla, bilateral dentate, and peri-dentate regions suggestive of focused areas of signal intensity [Figure 5]. No optic pathway glioma was seen.

The patient was kept on medical management for menorrhagia and no surgical intervention was advised.

DISCUSSION

We present the available reported cases of uterine plexiform neurofibromas, as shown in Table 1.

While Gwacham *et al.*[11] reported a case of localized plexiform neurofibroma of the uterus, Nalbantoğlu *et al.*[6] reported a case of pelvic plexiform neurofibroma causing mass effect on the uterus with no infiltration.

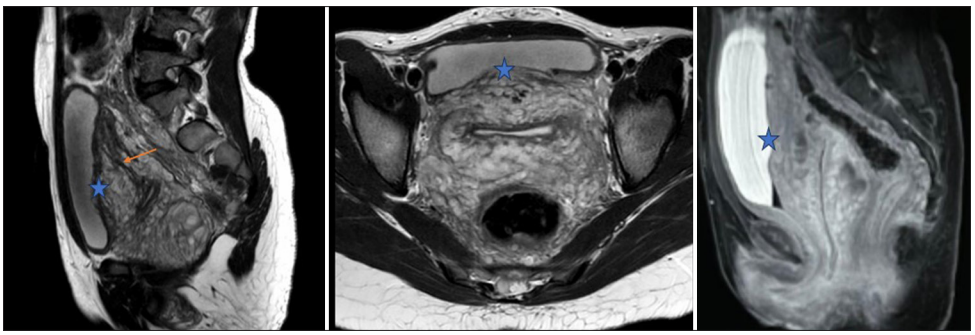

Figure 3: Sagittal and axial T2WI and post-contrast sagittal images show diffusely infiltrative plexiform neurofibroma-infiltrates myometrium with thinned-out endometrium as shown by the red arrow with infiltration and encasement of rectum and urethra. The posterior bladder wall is infiltrated as depicted by blue stars.

Table 1: Review of literature on genitourinary plexiform neurofibroma involving the uterus.

Article name	Age of patient	Complaints	Other features of NF1	Imaging findings
Gwacham *et al.*[11]	39/F	Heavy menstrual bleeding and abdominopelvic pain for 1 year	Known NF1	8×3.5 cm localised uterine plexiform neurofibroma
Nalbantoğlu *et al.*[6]	10/F	Painful and slowly progressive genital swelling.	Diagnosed NF1: 15 café-au-lait macules and multiple neurofibromas, lisch nodules	Ambiguous genitalia with clitoromegaly with plexiform neurofibroma involving pelvic floor, peri-pubic region, external genital region with normal pelvic organs
Mansour *et al.*[12]	3 case reports in girls aged 7, 9 and 17, respectively.	Presented with difficulty in micturition and defecation	Diagnosed NF1 in two of three patients	Pelvic masses encasing uterus, rectum and urinary bladder

NF1: Neurofibromatosis type 1

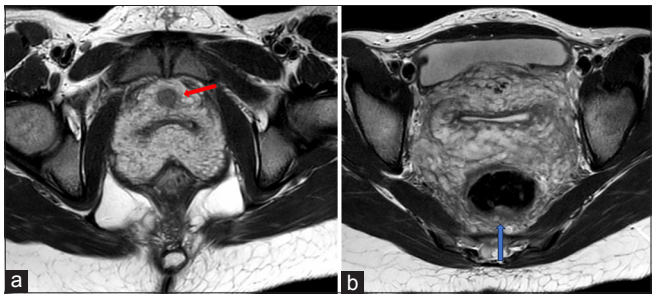

Figure 4: (a-b) Axial T2W images show hyperintense mass infiltrating the cervix and vagina and encasing the urethra (red arrow) and rectum (blue arrow).

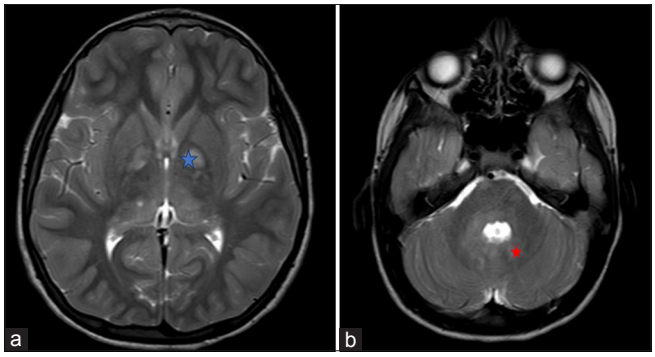

Figure 5: (a-b) Axial T2-weighted images of brains show hyperintensities in bilateral globi pallidi, thalami (blue star) and bilateral dentate and peridentate regions (red star) suggestive of foci of altered signal intensity (FASI) seen in neurofibromatosis.

Mansour *et al.*[12] reported cases similar to our case report with plexiform neurofibromas infiltrating the uterus and surrounding pelvic organs.

Differentials include vascular malformations in the pelvis which appear as serpentine flow voids with intense enhancement on post-contrast sequences and schwannomas (localized exophytic homogenously enhancing masses along nerve trunks).

CONCLUSION

Lymphangiomas, meningoceles, and teratomas are other differentials to be considered. Patients presenting with multiple café-au-lait spots and genital enlargement need to be screened for genitourinary tract neurofibromatosis.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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