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Case Report

Bladder plexiform neurofibroma in a 4-year-old male child with neurofibromatosis type 1: A case report

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Abstract

Bladder plexiform neurofibroma (PN) is a rare disease entity that is challenging to treat. We report a 4 year old male child who had presented with a suprapubic abdominal swelling and irritative voiding symptoms of one year duration. The child had a palpable suprapubic mass, multiple café au lait macules over his body. The father had similar skin lesions. Imaging revealed diffuse asymmetric thickening of the bladder wall. The diagnosis was confirmed by immunohistochemistry. Selumetinib was offered as a treatment option.

Key words: Bladder, plexiform, neurofibroma, child, Ethiopia

Introduction

Neurofibromatosis type 1 (NF-1) is a well-characterized autosomal dominant genetic disorder presenting with a diverse range of clinical manifestations. It affects roughly 1 in 2,600 to 1 in 3,000 individuals [1]. Plexiform neurofibromas are benign peripheral –nerve sheath tumors that occurs in up to 57 % of NF-1 pediatric cases [2]. Genitourinary involvement in NF1 is rare, with less than 80 cases reported in literature to date [1]. We report a 4 years old male child diagnosed to have a bladder plexiform neurofibroma, at the largest tertiary hospital in Ethiopia Tikur Anbessa Specialized Hospital (TASH). TASH receives referrals from government and private medical institutions, nationwide.

Case presentation

A 4-year-old male child presented with progressively increasing lower abdominal swelling, frequency, hesitancy, dysuria and hematuria of one year duration. The child had these voiding symptoms a few weeks prior to the abdominal swelling. The abdominal swelling started to rapidly increase in size one month prior to presentation at the institution. The child has

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no symptoms of urinary retention or previous catheterization.

The examination revealed a well-nourished and comfortable child with stable vital signs. He had a 4x4 cm suprapubic firm, non- tender palpable mass, multiple café au lait macules (CALMs) most greater than 5mm, particularly on his trunk, and a huge hyperpigmented patch on his right buttock (Figure 1, Supporting file). His father had CALMs as well. Complete blood count and urine analysis were non-revealing.

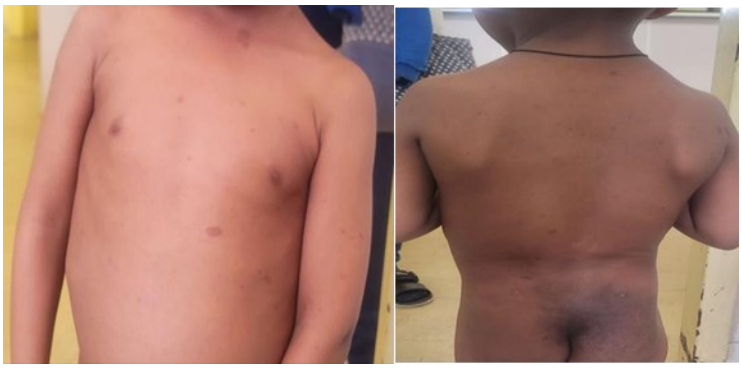

Figure 1: The presence of CALMs on the trunk.

The CT imaging demonstrated contrast enhancing diffuse asymmetric circumferential thickening of the urinary bladder wall measuring 2.2cm with evidence of mild bilateral pelvicalyceal dilatation. The renal parenchyma has normal thickness with symmetric contrast enhancement and excretion. The lumbosacral vertebral bones have normal CT findings (Figure 2 and 3, supporting files).

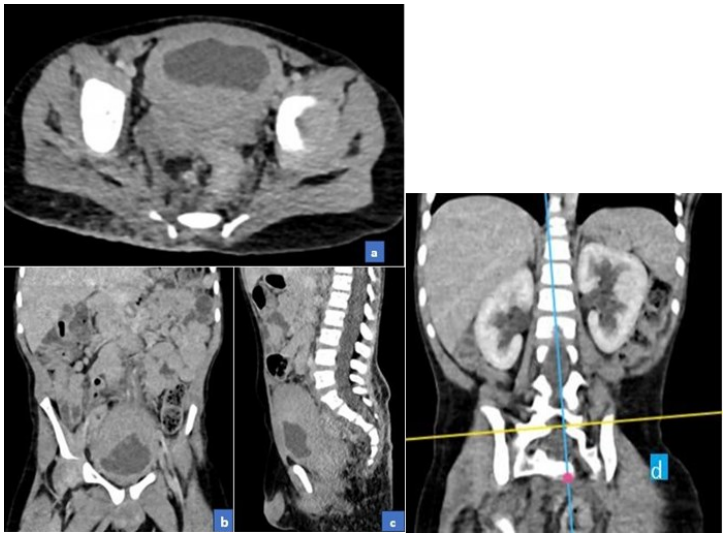

Figure 2: Post contrast abdominal-pelvic CT in axial (a), coronal (b) and sagittal (c) sections depicting homogenous diffuse asymmetric circumferential urinary bladder wall thickening. (d)Post contrast CT scan with coronal 3D MPR image depicting mild bilateral hydronephrosis

Gross specimen: one core needle fragment, 0.4x0.3cm.

Microscopic examination: small tissue composed of cellular nodules of myxoid degeneration and wavy spindle cells interspersed between skeletal muscle fragments forming a plexiform like architecture (Figure 3a and 3b, Supporting file).

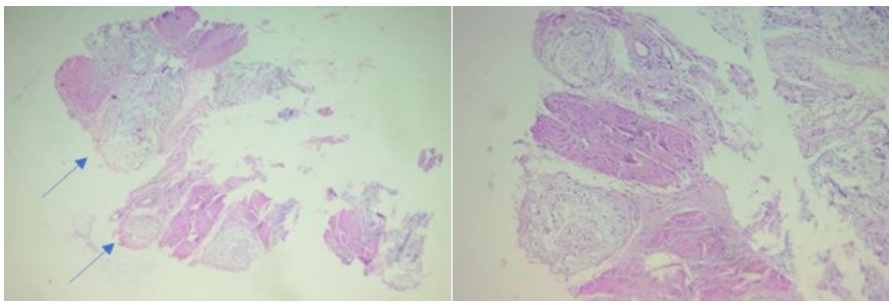

Figure 3a: Low Power (4x objective magnification) of submitted tissue showing plexiform like architecture (Arrow) B. 10x objective showing the schwann cells and myxoid stromal background.

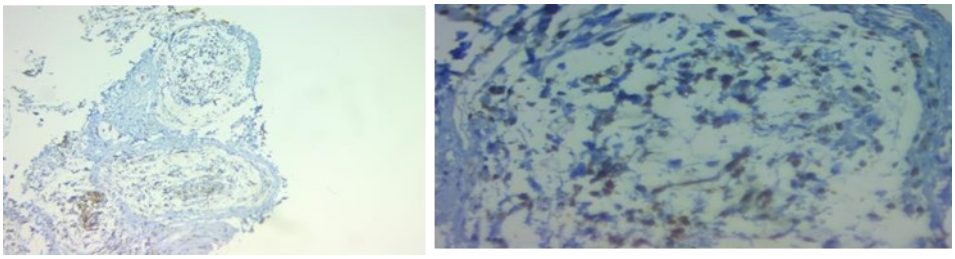

Figure 3b: S-100 stain showing cytoplasmic and nuclear staining in the schwann cells (10x objective) and B. 40x objective showing similar staining

Discussion

The presence of a bladder plexiform neurofibroma (BPN), CALMs in the child and the father fulfills the diagnosis of NF-1 [4]. Histology and immunohistochemistry have verified the diagnosis. Bladder involvement in NF1 is very rare and mostly affects the age group of 7 to 28 years with a ratio of 3:1 male predominance. It often arises from the nervous ganglia of the bladder wall, especially the vesicoprostatic plexus [9]. The clinical features of BPN include irritative voiding symptoms and hematuria due to recurrent urinary tract infections [5]. On imaging, BPN can manifest as a focal mass or as diffuse bladder wall thickening [3]. There is no definitive criteria for BPN. In contrast to our case, BPF without stigmata of NF-1 may be the initial presentation of NF – 1 [3,9]. The incidence of neurofibroma undergoing malignant transformation is 30% [10]. Bladder neurofibromatosis is almost always a benign process and malignancy has been reported in 5%–10% of the literature [9]. Radical cystectomy with urinary diversion is indicated for urinary obstruction with hydronephrosis or malignant transformation [5].

The diffuse and asymmetric nature of the bladder wall thickening makes our case inoperable. He was offered selumetinib, a MEK inhibitor. It works by inhibiting the molecule MEK, which plays a role in the Ras downstream signaling pathway and thus affects the cell proliferation in NF1-associated tumors. Gross et al reported 34 (68%) of 50 children with inoperable PNF had size reduction and improved symptoms. [8]. The child has not been started on treatment for financial reasons. Currently, he is on follow-up at the pediatric hematology/ oncology clinic. In conclusion, bladder PN should be suspected in the setting of irritative voiding symptoms, hematuria and bladder wall thickening, in the presence of NF-1. Inoperable BPN can benefit from selumetinib.

Declaration

Consent for publication

Verbal consent was obtained from the mother of the child and the mother also approved the publication of anonymized photo of the child. All personal data of the child were anonymized.

Conflict of interest

None to declare

Authors contribution

HA conceived the idea, compiled the child's history, physical findings, photo of the skin lesions and lab reports. AD involved in framing the idea, provided the pathology images and did the reporting. SS examined the imaging results did the radiology report. All authors involved in the write up of the manuscript, read the manuscript and approved the submission of manuscript.

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