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Case report

Early presentation of neurofibromatosis type I patient with clitoromegaly and café au lait spots: A case report ☆

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ABSTRACT

Neurofibromatosis is a rare genetic disorder that typically affects the nerves and causes benign tumors. It also affects different parts of the body, including the bone, skin, and genitourinary system. We report a case of a 6-year-old girl medically free who was referred to our institute with clitoromegaly and multiple café au lait spots on the skin. Clitoral mass excision was performed, and histopathology confirmed the diagnosis of clitoral plexiform neurofibroma as a primary presentation of Neurofibromatosis type I.

Introduction

Neurofibromatosis is a rare genetic disorder that causes benign tumors to develop that affect the brain, spinal cord, and nerves [1]. There are three types of neurofibromatosis: type I (NF1), type II (NF2), and Schwannomatosis. NF1, which was previously known as von Recklinghausen disease, is an autosomal dominant genetic disorder with multisystem involvement; it is the most common type of neurofibromatosis with an incidence of approximately 1 in 2,600-3,000 individuals [1]. Clinical manifestation of NF1 includes café au lait lesions, axillary or inguinal freckling, Lisch nodules, neurofibromas, and bone abnormalities. According to the National Institutes of Health in the United States, 2 or more of the following clinical features must be present for an NF1 diagnosis: six or more café au lait lesions, 2 or more neurofibromas, axillary or inguinal freckling, optic glioma, two or more Lisch nodules, bony lesions, or first-degree family history [2]. In some uncertain cases, genetic tests are performed to confirm the diagnosis. Genitourinary involvement in NF1 is rare, and the first report of clitoral neurofibroma was published by Haddad and Jones in 1960 [3].

Case report

A 6-year-old girl medically free presented at our institution, having been referred from a secondary hospital as a case of clitoromegaly at 4 years of age. For 2 years, the patient suffered from a clitoris mass along with multiple café au lait spots on

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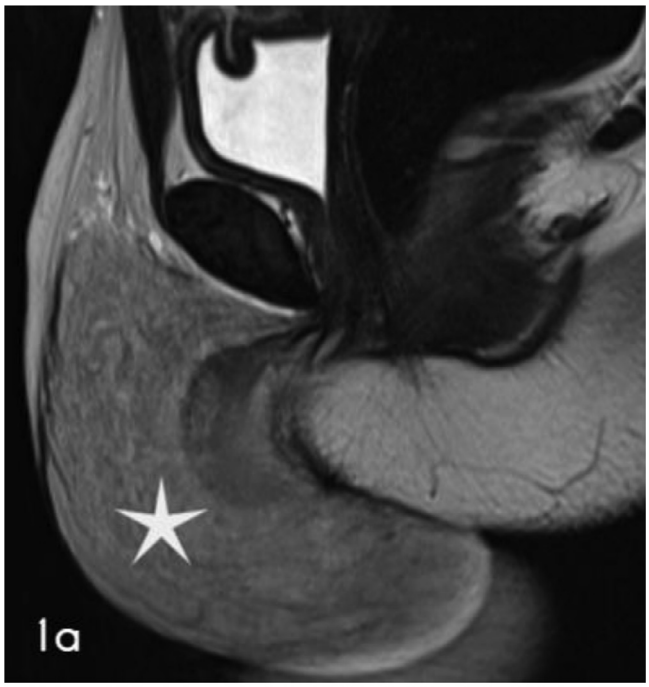

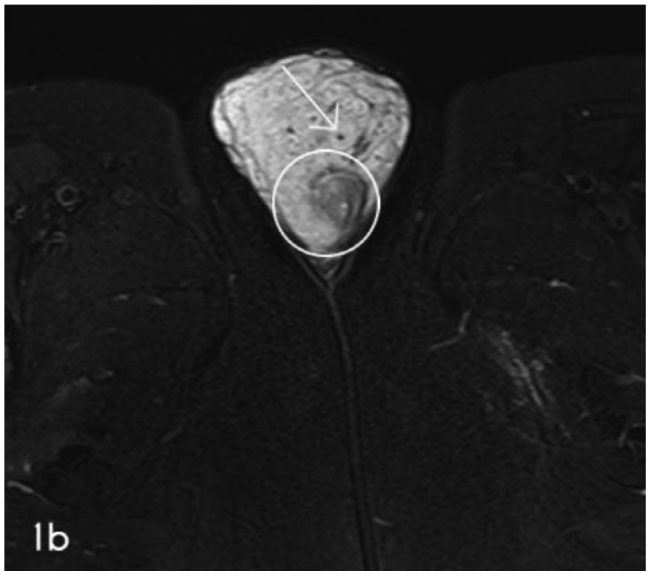

Fig. 1 – 1A and 1B: Sagittal T2 weighted images without fat saturation and axial T2 weighted images with fat saturation, demonstrating a large exophytic soft tissue mass centered at clitoral region (star) encasing the urethra (circle), with numerous tiny individual nodules predominantly moderately hyperintense on T2 containing numerous foci of central low signal intensity (arrow). It measures 9.4 x 4.6 x 3.7 cm in oblique CC, AP, and transverse dimensions respectively.

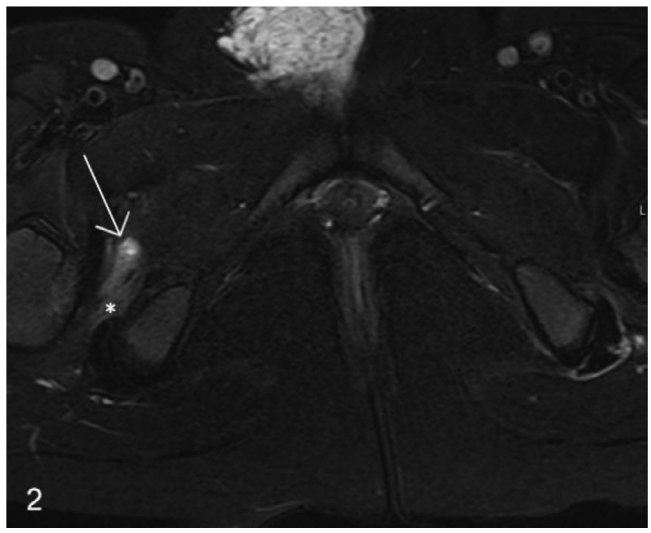

Fig. 2 – Axial T2 weighted images with fat saturation, demonstrating a similar appearance of a nodule along the thickened/edematous posterior branch of the obturator nerve (arrow), with signs of muscle denervation/reactive myositis involving the obturator internus muscle posteriorly (asterisk).

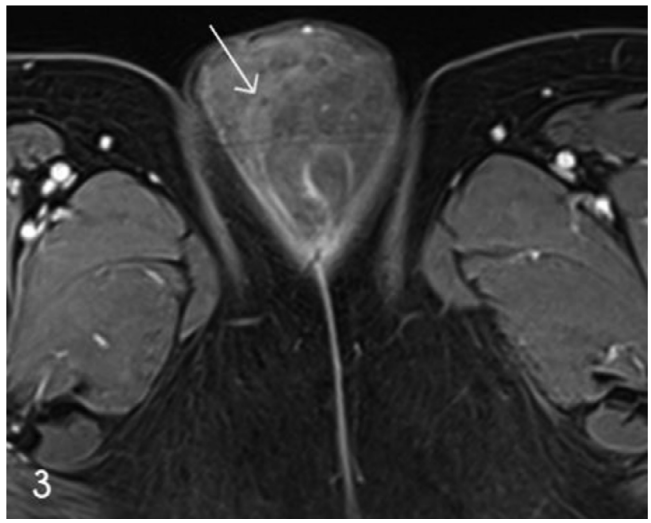

Fig. 3 – Axial post-gadolinium T1 weighted images showing mild heterogenous enhancement of the large exophytic clitoral soft tissue mass (arrow).

the skin of the abdomen and gluteal area. Results from genetic tests and endocrine workups were normal. The patient underwent cystoscopy and showed normal vaginal opening with annular hymen and normal urethral opening. She was diagnosed with NF1 using MRI. The karyotype indicated a normal female chromosome complement with no apparent abnormality (46, XX). Bone age assessment was performed for the patient, indicating the normal range for the patient's age.

Pelvic MRI revealed a large mass centered at the clitoris, which was consistent with a plexiform neurofibroma (Figs. 1A, 1B, 2, and 3). Additional small neurofibromas were identified along the posterior branch of the obturator nerve and surrounding the left ischial tuberosity. The remainder of the pelvic organs were unremarkable. Brain MRI showed focal areas of signal intensity in the right globus pallidus and mesial thalamus in the clinical context of NF1 (Fig. 4). Following the

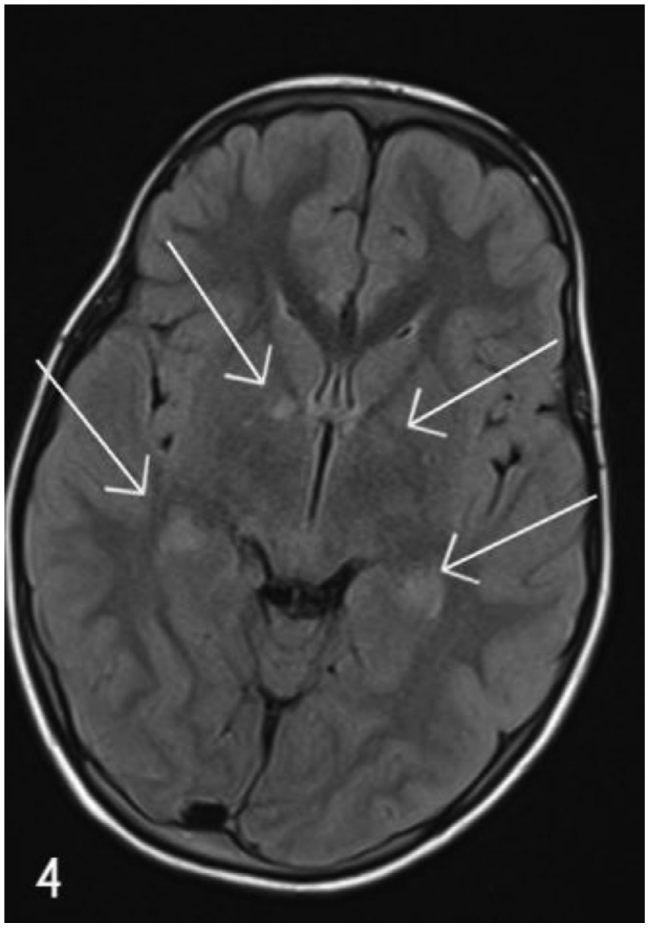

Fig. 4 – Axial FLAIR sequence of the brain, demonstrating foci of increased FLAIR signal intensity involving bilateral globus pallidus and hippocampus (arrows), most likely representing NF1-related focal areas of signal intensity (FASI).

diagnosis, the patient underwent clitoris mass excision. The surgical procedure was uneventful, and the patient was discharged to home the next day.

Discussion

In this case report, we describe clitoral involvement with plexiform neurofibroma in a child as a primary presentation of NF1. Despite advances in genetic testing, most NF1 cases are diagnosed based on clinical manifestations. NF1 patients typically present with café au lait lesions, axillary or inguinal freckling, Lisch nodules, neurofibromas, and bone abnormalities [2]. Clitoral involvement by neurofibromatosis is an extremely unusual presentation, and the incidence of clitoral neurofibroma is unknown. Some studies have reported that it is rare, accounting for 0.65% of all neurofibromas. The most frequent genital presentation of neurofibromatosis in females is clitoromegaly. The asymptomatic nature of clitoromegaly leads to delays in seeking medical attention, which might be a cause of underreporting [4,5]. The comorbidities of NF1 include disfiguring skin lesions, pain, visual or hearing impairment, seizures, and spinal cord compression complications. Despite the fact that some of these complications are uncommon, psychosocial stress is a significant problem [6]. It is rare for clitoral neurofibroma to progress into malignancy, but a case has been reported in which malignant Schwannomatosis occurred in connection with clitoral involvement [7]. Hence, MRI and histopathological evaluation are important in evaluating the disease. Genetic counseling is the first step in NF1 management. This is because if a parent is affected, the risk of neurofibromatosis for each pregnancy is 50%. Early diagnosis and surgical intervention play an important role in these patients. Surgical intervention is the main treatment, which typically involves clitoroplasty and preservation of the glandular tissue and neurovascular bundle [6].

Patient consent

Informed consent is not required from our Institutional Review Board due to the anonymized and retrospective nature of this report.

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