

Case Report

Plexiform neurofibromatosis of vulva

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ABSTRACT

Plexiform Neurofibromatosis of vulva is a rare, benign tumor of genital tract arising from nerve sheath of peripheral nerve. It may present as solitary lesion or as part of Von Recklinghausen's disease. Genitourinary neurofibroma is rare, however clitoris and labia majus happen to be the most frequent location of neurofibromatosis involving female genital tract. We present a case of plexiform neurofibroma arising from left labia majora in a young patient having features of Von Recklinghausen's disease. She complained of discomfort while walking due to rubbing of pedunculated mass arising from her labia majora. We carried out the surgical excision of the mass followed by primary repair of labia majora. She has been followed up on out-patient basis for about three months without any recurrence so far.

Keywords: Neurofibromatosis type 1, Plexiform neurofibroma of labia majora, Von Recklinghausen's disease

INTRODUCTION

Neurofibromatosis type 1 (NF1) also known as peripheral neurofibromatosis or Von Recklinghausen's disease is a systemic disorder of autosomal dominant inheritance. It affects all races and affects both genders equally, incidence being one in every 3000 of the population.¹ Important features of this disease include presence of hyper-pigmented skin lesions (café-au-lait spots), neurofibromas, iris hamartomas, macrocephaly, central nervous system tumors, defects of the skull and facial bones, and vascular lesions.² Genitourinary neurofibroma is rare with fewer than 40 paediatric cases of genitourinary neurofibromatosis reported in the literature. Neurofibroma involving only female genital tract is extremely uncommon.³ Among the genitourinary neurofibromas, vulva is the most commonly affected genital organ, whereas involvement of the uterine cervix, endometrium and ovaries have been very rarely reported.⁴ It may occur as solitary or multiple lesions as part of Von Recklinghausen's disease. We report a case of plexiform neurofibromatosis affecting labia majora in a 15 years old school girl.

CASE REPORT

AB, 15 year old short statured female patient reported to our surgical out-patient department with history of overhanging mass affecting her vulva since early childhood. Initially the mass was small in size and painless, but progressively increased in size during last three years, resulting in discomfort while walking as the mass used to rub against her inner thighs while walking. There was no history of trauma or bleeding per vagina. General physical examination revealed presence of multiple Café-au-lait spots over her trunk, size of spots being more than 15mm. Besides, she had multiple sessile neurofibromas over anterior chest wall and right eyebrow (Figure 1, 2). Ophthalmological consultation revealed presence of Lisch nodules over iris. Local examination showed a large pedunculated mass arising from the left labia majora (Figure 3). Mass was 10cm x 7cm in dimension and 5cm at the base with no involvement of labia minora and clitoris. There was no extension of the mass to the vagina, cervix or uterus and mass was non tender. Right labia majora and minora were normal. MRI exam (Figure 4) of perineum revealed overhanging mass

from left labia majora. Patient was subjected to surgical excision of mass and primary repair of labia majora under general anesthesia. Histopathological exam of excised specimen revealed spindle shaped cells with wavy nuclei arranged in a loose myxomatous stroma (Figure 5). Patient has been followed up for last three months and there has been no evidence of recurrence (Figure 6).



Figure 1: Neurofibroma affecting right eyebrow.

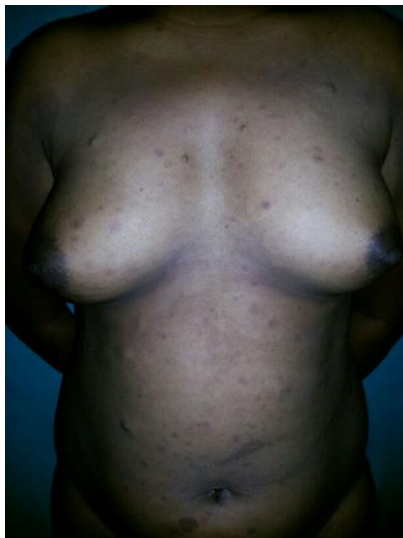


Figure 2: Multiple Café-au-lait spots and neurofibromas over her trunk.



Figure 3: Pedunculated overhanging mass affecting labia majora.

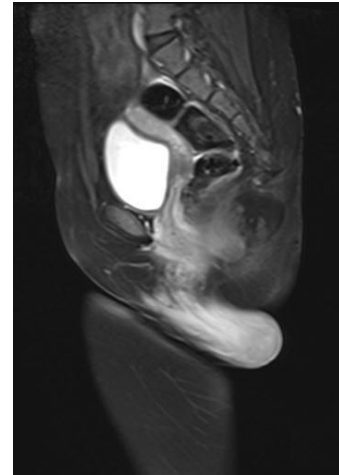


Figure 4: MRI perineum showing large well circumscribed exophytic lesion along left labia majora, with no extension into the vagina.

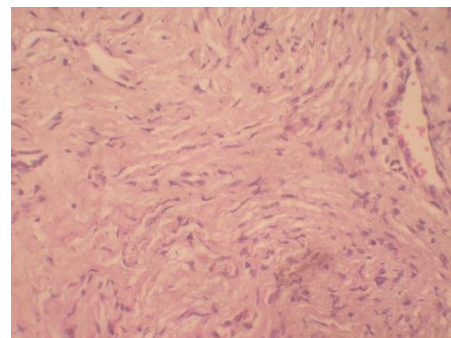


Figure 5: HP exam of excised mass showing spindle cells and nerve fibres with vascular channels in myxomatous stroma.



Figure 6: post op picture after three months showing nicely healed scar and no recurrence.

DISCUSSION

Neurofibromatosis is a hereditary neurologic disorder firstly described by the German pathologist Frederick Von Recklinghausen in 1882. Type 1 Neurofibromatosis (NF1)-or Von Recklinghausen's disease is transmitted on chromosome 17 and is mostly caused by mutation of NF1 gene whereas Type 2 neurofibromatosis (NF2) results mainly from a mutation of NF2 gene and transmitted on chromosome 33.

Vulval involvement is found in about 18% of women suffering from Von Recklinghausen's disease while

approximately 50% of all vulval neurofibromas are found in women with neurofibromatosis. Neurofibromas involving the female genital tract commonly involve the clitoris and the labia but may also affect the vagina, cervix endometrium, myometrium, and ovary and may also be associated with urinary tract neurofibromatosis.⁵

Genitourinary neurofibromas with clitoral involvement in neurofibromatosis type 1 are rare, and even more infrequent are the neurofibromas involving genitalia in males. The most frequent presenting sign of neurofibroma in females is clitoromegaly with pseudopenis. Labium majus neurofibroma not associated with clitoral involvement is extremely rare,⁶ like the present case being reported here.

The diagnosis of NF1 is largely based on clinical criteria established by the National Institutes of Health Consensus Development Conference i.e the presence of two or more of the following: the presence of six or more café-au-lait macules or neurofibromas, Lisch nodules, axillary or inguinal freckling, optic glioma, distinctive osseous lesions or first-degree relatives with NF1.⁷ 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.

Most common presenting symptoms of vulval neurofibromas are dyspareunia and chronic pelvic pain. Diagnostic imaging is very important to evaluate the distinction between superficial and invasive tumors, and MR is the technique of choice in planning surgical resection of the lesion. Individuals affected by NF1 harbor an increased risk for both benign and malignant tumors. Malignant transformation is usually observed in the form of neurosarcoma, incidence mentioned in literature being 7 to 13%.⁸

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Ethical approval: Not required

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