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A CASE REPORT OF LABIAL AND FACIAL NEUROFIBROMAS AS PRESENTING SYMPTOMS OF NEUROFIBROMATOSIS.

General Surgery

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KEYWORDS

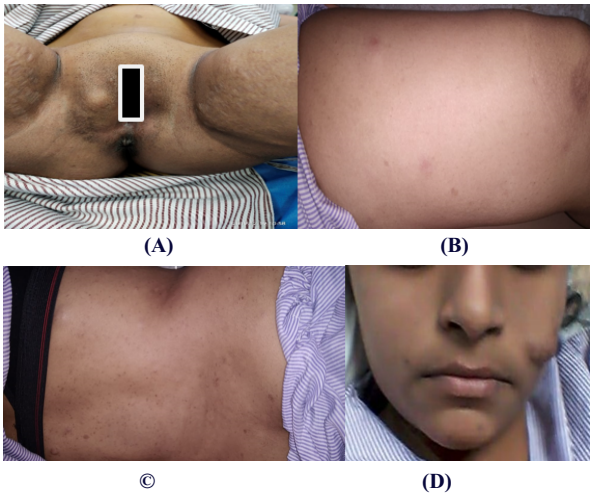
INTRODUCTION

Neurofibromatosis (NF) type-1 is an autosomal dominant neurocutaneous disorder also known as peripheral neurofibromatosis or Von Recklinghausen's disease, characterised by peripheral nerve sheath tumor (neurofibroma), hyper-pigmented skin lesions (cafe-au-lait spots), inter-triginous freckles, lisch nodules, macrocephaly, defects of skull and facial bones. Genital NF is extremely rare, Vulva being the most common type in it, and is often confused with hyper androgenic disorders resulting in unnecessary evaluations. Neurofibromas involving the orbital, temporal and facial regions are known as orbito-temporal neurofibromatosis (OTNF) seen in <1 %. It affects irrespective of race or gender. The incidence of this syndrome is 1 in 3500 individuals. Here we report a case of labial and facial neurofibroma in 18-year-old female.

CASE REPORT

A 18-year-old female came with complain of right labial and left cheek swelling since last 10 years, which increased in size gradually, associated with pain on movement. No history of fever or discharge from swelling.

On examination, a 3*2 cm single, soft, compressible swelling, having ill-defined margin was present on left cheek. Another 4*3 cm single, firm, freely mobile swelling with well-defined margin was present over right labia. Few café-au-lait lesions were present over the back and thigh. No history of hearing or visual disturbances, convulsions or dizziness was present.



(A) (B)

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Figure 1: Different Manifestations Of Neurofibromatosis

(a) Labial Neurofibroma; (b) (c) Cafe-au-lait Spots; (d) Facial Neurofibroma

All blood investigations including hormonal investigations were found to be within normal limits. **Ultrasonography** was suggestive of 23*6 mm sized cystic lesion, showing multiple neovessels with echogenic component was noted over left cheek arterial and venous waveform were identified in vessels, suggestive of vascular malformation over left cheek. Another approximately 35*13 mm sized, oval shaped, hypo echoic lesion was seen in right labia, with central vessel showing arterial flow, p/o right labial testes. **MRI** was suggestive of 35*11 mm sized well defined, oval shaped with altered signal intensity lesion, which appear hypointense on T1, isotense with hyperintense rim on T2, in right labia with surrounding subcutaneous oedema. Well defined, oval shaped altered signal intensity lesion in right labia possibility of right labial testes likely.

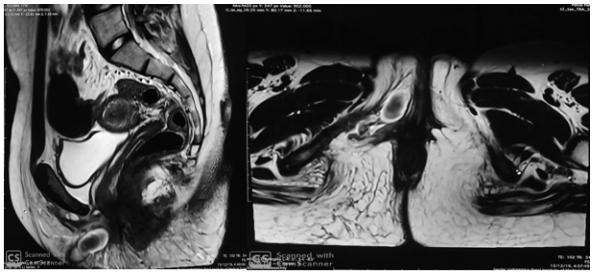

Figure 2: MRI Pelvis

Patient was operated for swelling **Excision and Biopsy**. A single, soft consistency, whitish tissue swelling of 3*2 cm was excised from left cheek and sent for Histopathological examination. Similarly, the Labial swelling was excised from the mucocutaneous junction; which was approximately 4*2 cm in size, single, and firm in consistency and sent for histopathological examination. Both the biopsy reports were suggestive of neurofibroma.

The patient was discharged on 5th post-operative day with healthy stich line.

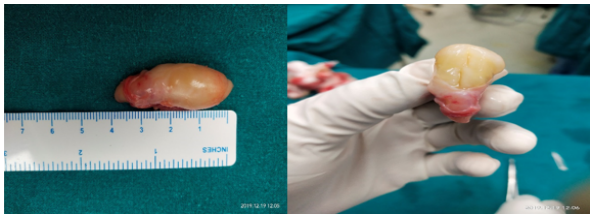

Figure 3: Excised Specimen Of Labial Neurofibroma

DISCUSSION

NF are of three types are Neurofibromatosis type I (Nf1), Neurofibromatosis type II (NF2), and Schwannomatosis.

Neurofibromatosis type I is the most common inherited disorder caused by a mutation of a gene on the long arm of chromosome 17. [2] Neurofibromatosis type I gene encodes a protein known as neurofibromin which acts as a tumor suppressor protein, an in activator of Ras. It is an autosomal dominant genetic disorder .The incidence of NF-1 is about 1 in 3500 live births. [3] Neurofibromas can be classified according to their anatomical location: -Dermal, intra-neural, and plexiform. Dermal neurofibromas are associated with a single peripheral nerve, while plexiform neurofibromas are associated with multiple nerve bundles. Plexiform neurofibromas are more difficult to treat and can transform into malignant tumors. Dermal neurofibroma does not become malignant.

Diagnosis	NF 1	NF2
Gene mutation	NF1 tumor suppressor gene, codes the protein neurofibromin	NF2 tumor suppressor gene, codes the protein merlin
Location of mutated gene	Chromosome 17	Chromosome 22
Main clinical features	Café-au-lait-spots Multiple neurofibromas Lisch nodules	Bilateral acoustic neuromas

Diagnostic criteria for NF1(4) (NIH has seven diagnostic criteria for neurofibromatosis type 1. At least 2 of the clinical features must be present to make the diagnosis)
1. Six or more cafe-au-lait spots, >5 mm in diameter in pre-pubertal and > 15 mm in post-pubertal individuals.
2. Two or more neurofibromas or one or more plexiform neurofibroma
3. Axillary or groin freckling
4. Optic Glioma
5. Two or more Lisch nodules
6. Sphenoid dysplasia, dysplasia or thinning of long bone cortex
7. First-degree relative with neurofibromatosis type 1.

Cutaneous manifestation and common age of onset for patients with Neurofibromatosis type 1

Cutaneous manifestations	Age of onset
Cafe-au-lait spots	Birth to age 12
Axillary freckling and lisch nodules	Age 3 to adolescence
Subcutaneous and cutaneous neurofibroma	Adolescence

Neurofibroma of genital tract is very uncommon. Amongst them vulva is the most frequent location of neurofibroma followed by clitoris and labia but rarely found in vagina, cervix, endometrium, myometrium as well as urinary tract (5).

Medical complications

Dermal neurofibromas can lead to stinging, itching, pain, and disfiguration.

Plexiform neurofibroma can cause disfigurement, neurological, and can transform into a malignant peripheral nerve sheath tumor.[5]

TREATMENT

Dermal neurofibroma

Dermal neurofibromas are not usually surgically removed unless they are painful or disfiguring, because there are generally so many of them and they are not dangerous.

Plexiform neurofibroma

Removal of plexiform neurofibromas is difficult because they can be large and cross tissue boundaries. However, besides pain, plexiform neurofibromas are sometimes removed due to the possibility of malignant transformation.

Medications

ACE inhibitors work by indirectly down regulating TGF-beta, which is a growth factor that has been shown to influence the development of tumors.[18]

CONCLUSION

In conclusion, despite its rarity, physicians should consider neurofibroma as a differential diagnosis of a vulval swelling, especially in patients who have other clinical manifestations of neurofibromatosis, or with family history of the disease, that might help in the management by wide excision of the mass with a safety margin at the primary surgery to prevent its recurrence

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