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CASE REPORT **OPEN ACCESS**

A rare case report on Neurofibromatosis type 7

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ABSTRACT
A 65 year old presented to dermatology OPD with multiple neurofirbromas with onset two years back without café au lait spots or lisch nodules. Patient did not have any family history of similar lesions. Skin biopsy confirmed the diagnosis of neurofibromatosis. Neurofibromatosis type 3 to type 8 is very rare. Hence, this is a case of late adult onset neurofibromatosis type 7 which is very rare in occurrence.
Key words : *Neurofibromatosis type 7 **(**NF-7), **A**dult onset neurofibromatosis*

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INTRODUCTION

Neurofibromatosis (NF) is an autosomal dominant condition. Neurofibromatosis is also known as neurocutaneous syndrome.[1] NF classification system proposed by Riccardi classifies NF into 8 types.[2] Most types of NF present with neurofibroma, axillary freckles and café-au-lait spots. NF type 1 (NF-1, also termed as von Recklinghausen disease) is the commonest neurofibromatosis and NF type 2 (NF-2) is second most common neurofibromatosis. Most neurofibromas are benign, although they may become malignant in some cases. Neurofibromas appear after 3rd decade of life and are generally not associated with café-au-lait spots. NF type 7 is very rare with a presentation that is very atypical.[1] Hence, we are presenting a case report on NF-7.

CASE REPORT

A 65 year old male patient came to the hospital with chief complaint of multiple elevated skin coloured lesions present since two years not associated with itching or pain. A single lesion appeared over the nose two years back which increased in size and similar lesions developed over face, chest, abdomen, back, forearms and arms.
On examination, multiple cutaneous nodules of varying sizes, symmetrical in distribution were present over face, chest, abdomen, back, upper limbs and few over thighs. Button hole sign was absent (Figure 1). A single fluctuant, non tender, skin coloured swelling was present over the right middle finger.
Other cutaneous findings like café-au-lait macules and freckles were absent. Patient did not have difficulty in hearing, tinnitus or vertigo. Patient did not have any weakness in extremities. Family history of neurofibroma was absent. Slit lamp examination did not reveal any lisch nodules. On biopsy, loose proliferation of spindle cells were found in superficial epidermis compatible with neurofibroma (Figure 2).

DISCUSSION

NF is an inherited autosomal dominant disorder.[2] NF-1 is due to the gene neurofibromin located on 17q11 and NF-2 is due to the gene schwannomin or merlin located on 22q12.[1] NF classification system, which divides NF into eight types was proposed by Riccardi.[3] According to NIH, diagnostic criteria of NF1 is: minimum 6 café au lait spots with a minimum size of 0.5 cm before onset of puberty or a minimum diameter of size 1.5 cm after onset of puberty, inguinal as well as axillary freckling, minimum 2 Lisch nodules, optic glioma, bony lesions like dysplasia of sphenoid wing and first degree family history with NF1.[4] In NF2 there is a 50% of autosomal dominance inheritance and 50% chances of sporadic mutation.[5] NF-2 diagnosis requires at least one of these following features: (1) eighth nerve masses on both side, (2) positive family history with NF-2 with eighth nerve mass present unilaterally, or any two of

the mentioned lesions: neurofibroma, meningialtumour, glioma, schwann cell tumor, or juvenile posterior subcapsular lenticular opacity. [1] NF-3 (also referred to as mixed form) consisting of multiple tumors of brain and spinal cord slong with café-au-lait spots and neurofibromas, NF type 4 (variant form) which consists of both neurofibromas and café-au-lait spots but café-au-lait spots may disappear later, NF type 5 (NF-5: segmental form) in which segmental café-au-lait spots and/ or neurofibromas are present that is limited to a segmental distribution, NF type 6 presents with only café-au-lait spots [3]. NF type 7 is a late-onset form in which neurofibromatous lesions appear after third decade of life and café-au-lait macules are not seen.[1]Unspecified form, or NF-8, does not match the criteria of any other category. NF-7 is a rare varient of NF wherein schwannoma or neurofibroma, skin freckling occurs later in life but lack café-au-lait macule. [3] Probably the first case of NF7 was reported in 1941, when a case of paraplegia aged 65 years was operated and was found to have neurofibroma of the spinal cord. Later, he developed multiple sessile neurofibromas all over body.[3] Here, we report a case of neurofibromatosis speculating as NF7.

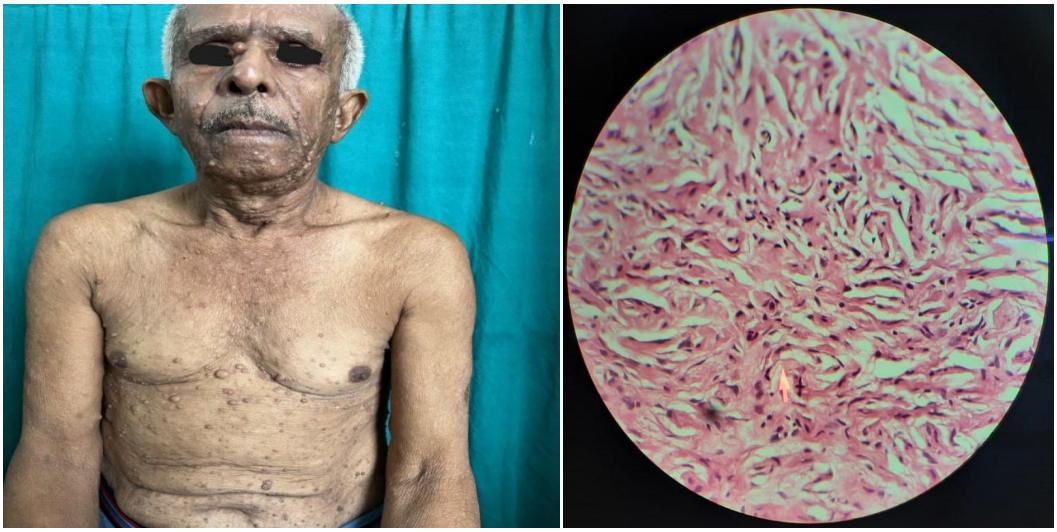

Figure 1. Multiple cutaneous nodules of varying sizes, symmetrical in distribution were present over face, chest, and abdomen

Figure 2. Loose proliferation of spindle cells found in superficial epidermis compatible with neurofibroma on H & E Stain (10X40)

CONCLUSION

Neurofibromatosis type 7 (NF-7) is an extremely rare form of the disorder that often manifests in middle age or later and is defined by the presence of neurofibromas or shwannomas but not café-au-lait spots. Due to the rarity of NF-7 in the literature, we report a case of NF-7.

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