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ISOLATED GIANT NEUROFIBROMA: A UNIQUE PRESENTATION

Pankaj Gupta[1], Anil Kumar Saroj*[2], Varsha Kumar[3]

[1]Resident, [2]Assistant Professor, P.G. Department of Surgery, M.L.N. Medical College, Allahabad, Uttar Pradesh – 211001, India.
[3]Department of Pathology, M.L.N. Medical College, Allahabad, Uttar Pradesh – 211001, India.

Corresponding Author:- **Anil Kumar Saroj**
E-mail: dranilsaroj123@gmail.com

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ABSTRACT
Neurofibroma is a benign nerve sheath tumor of the peripheral nervous system. It is usually found in individuals with neurofibromatosis type 1 (NF1), a genetically-inherited disease. This can result in a range of symptoms from physical disfigurement, pain to cognitive disability. To the best of our knowledge, this case corresponds to Joseph Merrick, famous as *Elephant Man of 19th century* and *Tree man of Java*.

INTRODUCTION

Neurofibroma arises from Schwann cells that exhibit bi-allelic inactivation of NF1 gene, which codes for the neurofibromin protein [1]. This protein is responsible for regulation of RAS mediated cell growth pathway. NF1 gene is composed of 60 axons spanning 350kb of genomic data, and maps to chromosomal region *17qll.2* [1,2] .Neurofibromas have been subdivided into two broad categories, dermal neurofibroma and plexiform neurofibroma. According to the World Health Organization classification system, dermal and plexiform neurofibromas are grade I tumors [3].

Dermal neurofibromas, also referred as cutaneous neurofibromas, are associated with a single peripheral nerve bundle involvement. They typically arise in the teenage, associated with onset of puberty. They continue to increase in number and size throughout adulthood. They look like small swellings on or under the skin. While dermal neurofibromas can lead to stinging, itching, pain and disfiguration, there is no evidence of malignant transformation [3]. Plexiform neurofibromas are often congenital, associated with multiple nerve bundle involvement and more troublesome [3,5]. They can be very large and can cause pain, disfigurement, neurological and other clinical deficits. Internal plexiform neurofibromas are very difficult to remove completely because they extend through multiple layers of tissue and the attempt to remove may damage healthy tissue or organs.[5] About 10% of plexiform neurofibroma may undergo malignant transformation converting into malignant peripheral nerve sheath tumour (MPNST) [3,4].

CASE SUMMARY

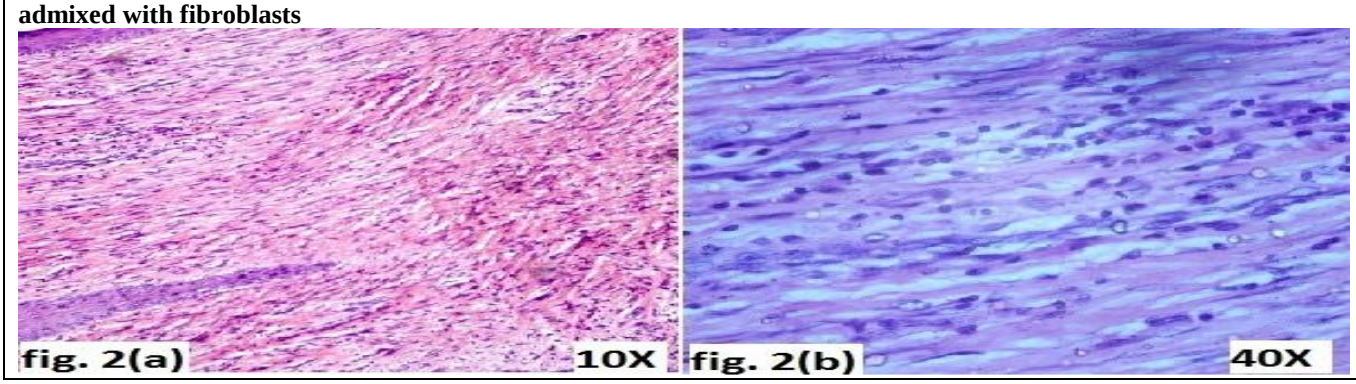
A 30 years old male patient presented with complaints of slowly progressive fungating mass over left foot and ankle region for last 8 years. On clinical examination, a firm, globular, non-tender, keratinizing growth of football size (30X30cm.) was present over the left ankle region, covering dorsum of foot and lower one third of leg. It was nonmobile, fixed to underlying structures as well as to overlying skin with focal areas of haemorrhage and pinkish discoloration. Surface was irregular cauliflower like with well-defined margins. It was associated with dark brown pigmentation of left lower limb. Neurological examination was normal and no sign of

neurocutaneous markers was present. Routine blood investigations were unremarkable. Patient underwent wide given over the mass and deepened to define the plane between mass and normal subcutaneous tissue. The tumour dissected off *(in toto)* from the subcutaneous tissue plane alongwith overlying skin. Haemostasis was achieved and sterile compression dressing with limb elevation given. surgical excision of the mass. A longitudinal incision was

The excised tumour was of 24 lb by weight. On 15th postoperative day, split skin grafting of the wound was done and patient discharged after one week. Specimen was sent for histopathological examination was suggestive of neurofibroma.

Figure 1. (a) showing neurofibroma of left ankle region involving dorsum of foot and lower one third of leg with focal areas of haemorrhage and pinkish discoloration, (b) excised tumour (24 lb by weight), (c) Post-operative status of the limb after split skin grafting

Figure 2 (a,b). photomicrograph showing proliferation of neural cells having elongated nuclei with pointed ends admixed with fibroblasts



DISCUSSION

The prototype presentation of an isolated giant neurofibroma is quiet rare. Neurofibromatosis type 1 also known as, Von Recklinghausen's Disease, is inherited as autosomal dominant trait.[3] NF1 is associated with presence of neurofibromas, café-au-lait macules, axillary freckles, lisch nodules over iris and optic glioma.[3,4] Neurofibromas are commonly treated with surgical removal. Dermal neurofibromas are not usually removed unless they are painful or disfiguring, because there are generally numerous and benign in nature (no malignant transformation).[5] Removal of plexiform neurofibromas can be more difficult because they can be larger and cross tissue boundaries. They are always removed due to the possibility of malignant transformation.[4,5] Sometimes plexiform neurofibromas are found at rare locations, as urinary bladder and left ventricular mitral valve.[5] Other treatment options are radiationand chemotherapy. However, radiation is generally not used as a treatment for plexiform neurofibromas because of risk of malignant transformation. Carbon dioxide lasers have been used to remove dermal neurofibromas.

However, it is strongly recommended that a test therapy should be performed before, to judge the effectiveness of the procedure and acceptability of scar. ACE inhibitors act indirectly by down regulating TGF-beta receptors, which is a growth factor responsible for development of tumor, therefore ACE inhibitors have been proposed as a novel future treatment option of neurofibroma.[6]

CONCLUSION

This prototype presentation of an isolated giant neurofibroma is quiet rare. This gentleman could be regarded as *Elephant man of India*. Before considering the surgical resection, all patients should be screened for possibility of neurofibromatosis. Wide local excision is done in symptomatic cases and radiotherapy is contraindicated due to remote risk of malignant transformation.

Conflicts of interest: None declared.

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