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Case Report

Giant neurofibroma around the knee in a 34-year-old male: a case report

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ABSTRACT

Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder characterized by multiple neurofibromas, café-au-lait macules, and skeletal abnormalities. Plexiform neurofibromas can attain large sizes, causing functional impairment and cosmetic deformity. Surgical excision remains the primary treatment for symptomatic lesions. A 34-year-old male with a known history of NF1 presented to the General Surgery OPD with a progressively enlarging mass around the left knee and multiple swellings over the body for 15 years. The swelling around left knee was associated with discomfort during walking and restriction of knee movements. Physical examination revealed a large, soft-to-firm, non-tender mass measuring approximately 10×12 cm involving the anterior aspect of the knee. Magnetic resonance imaging (MRI) of left knee suggested a large soft tissue mass consistent with neurofibroma without intra-articular extension. CECT Chest suggested few apical bullas and multiple cutaneous fibromas. The patient underwent complete surgical excision of the mass under general anaesthesia. Histopathological examination confirmed the diagnosis of neurofibroma. The postoperative period was uneventful, and the patient showed significant functional improvement during follow-up. Giant neurofibromas around the knee are uncommon and can significantly impair mobility. Early diagnosis and complete surgical excision can provide symptomatic relief, improve function, and prevent further complications.

Keywords: Neurofibromatosis type 1, Neurofibroma, Knee mass, Surgical excision, Peripheral nerve sheath tumor, Case report

INTRODUCTION

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is one of the most common inherited neurocutaneous disorders, affecting approximately 1 in 3,000 individuals worldwide. It is caused by pathogenic variants in the NF1 gene located on chromosome 17 and follows an autosomal dominant pattern of inheritance.[1,2] The disorder is characterized by the development of multiple neurofibromas, café-au-lait macules, axillary freckling, Lisch nodules, skeletal abnormalities, and various neurological manifestations.[1-4]

Neurofibromas are benign peripheral nerve sheath tumors that may occur as localized, diffuse, or plexiform lesions. Plexiform neurofibromas are considered pathognomonic for NF1 and can enlarge progressively, leading to pain, functional disability, cosmetic disfigurement, and, in some cases, malignant transformation into malignant peripheral nerve sheath tumors (MPNSTs).[5,6] Recent advances in the understanding of NF1 biology have led to the development of targeted therapies such as MEK inhibitors; however, surgical excision remains the standard treatment for symptomatic, enlarging, or functionally disabling lesions.[6,7]

Current international consensus guidelines emphasize multidisciplinary evaluation, imaging assessment, and individualized treatment planning for patients with NF1-associated tumors.[8] Histopathological examination remains essential for confirming diagnosis and excluding malignant transformation according to the latest WHO classification of soft tissue tumors.[9] We present a case of a giant neurofibroma around the knee in a 34-year-old male with NF1 that was successfully managed by complete surgical excision

CASE REPORT

A 34-year-old male presented to the General Surgery Department, RKDF Hospital, with complaints of a gradually enlarging swelling around his left knee for approximately 15 years. The patient reported progressive increase in size of the mass associated with difficulty in walking, discomfort during prolonged standing, and restriction of knee flexion (Figure 1 and 2).

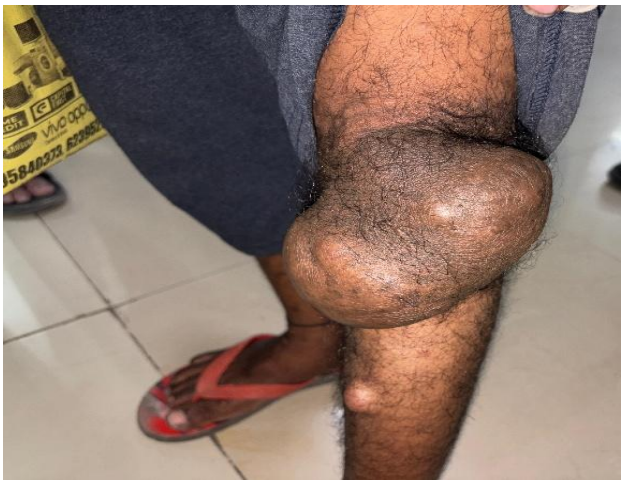

Figure 1: Preoperative clinical photograph showing giant neurofibroma over the left knee.

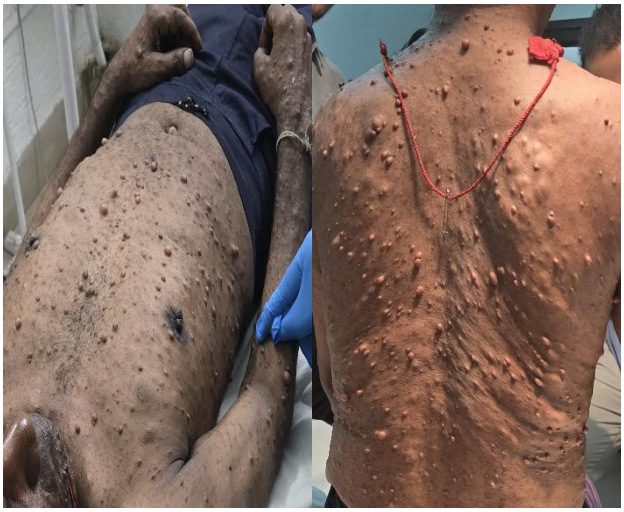

Figure 2: Preoperative clinical photograph showing giant neurofibroma over the body.

The patient was a known case of NF1, diagnosed during adolescence based on multiple cutaneous neurofibromas and café-au-lait macules. There was no history of trauma, ulceration, discharge, fever, or weight loss.

Clinical examination

General examination revealed multiple cutaneous neurofibromas distributed over the trunk and upper limbs along with multiple café-au-lait spots.

Local examination of the left knee showed: Large soft-to-firm swelling over the medial and anterior aspect of the knee. Measured approximately: 10×12 cm. Non-tender and irregular surface. Skin over the swelling was stretched but intact. No local rise in temperature. Knee movements were mildly restricted due to the mass effect. Distal neurovascular status was normal.

Investigations

Routine hematological and biochemical investigations were within normal limits.

MRI of the knee demonstrated a well-defined heterogeneous soft tissue mass arising from peripheral nerve structures around the knee joint without evidence of bony involvement or malignant transformation.

Surgical management

After preoperative evaluation and informed consent, the patient was planned for surgical excision under general anaesthesia.

An elliptical incision was made over the swelling. Careful dissection was performed to identify and preserve adjacent neurovascular structures. The lesion was well circumscribed and encapsulated and was excised completely. Hemostasis was secured, and the wound was closed in layers (Figure 3).

The excised specimen measured approximately 10×12×8 cm and was sent for histopathological examination (Figure 4).

Histopathological findings

Microscopic examination revealed spindle-shaped cells with wavy nuclei embedded in a collagenous and myxoid stroma, consistent with neurofibroma. No evidence of atypia, necrosis, or malignancy was identified.

Postoperative course

The postoperative period was uneventful. The patient was discharged on the sixth postoperative day. At follow-up after one month, the wound had healed well, and the patient reported significant improvement in walking

ability and knee mobility. No recurrence was observed during subsequent follow-up visits.

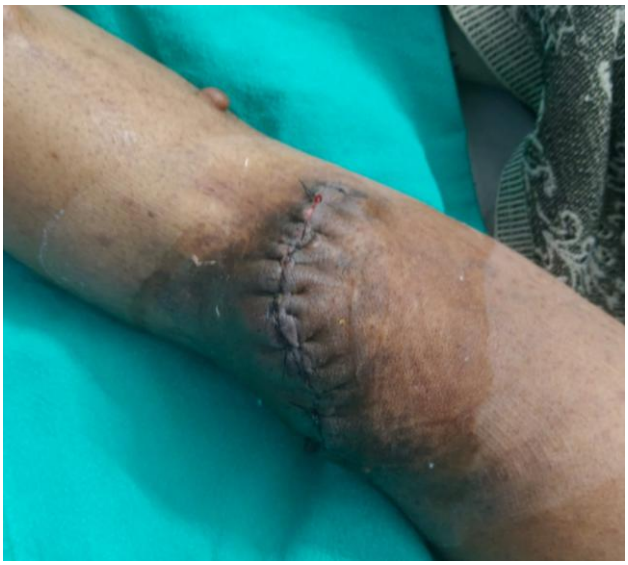

Figure 3: Intraoperative photograph showing complete excision of the neurofibroma.

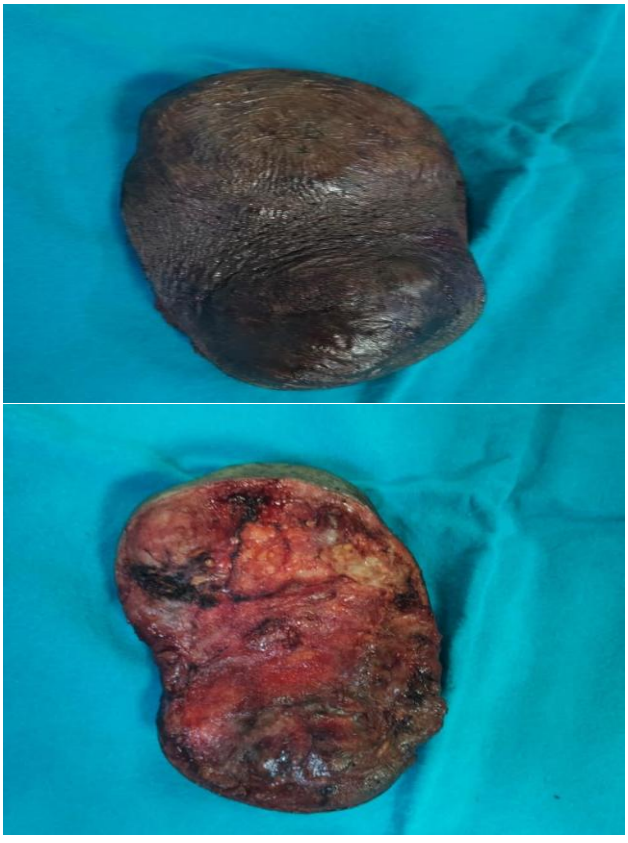

Figure 4: Excised specimen measuring approximately 10×12×8 cm.

DISCUSSION

Neurofibromas are benign tumors arising from Schwann cells, fibroblasts, perineural cells, and mast cells within peripheral nerve sheaths and are among the most common manifestations of NF1.[1,2] While cutaneous neurofibromas are frequently encountered, giant localized neurofibromas involving the knee region are uncommon and may significantly affect mobility and quality of life.[3,5]

Plexiform and large localized neurofibromas often exhibit progressive growth and may cause pain, joint restriction, cosmetic deformity, and compression of adjacent structures.[5,10] Imaging studies, particularly MRI, play a critical role in defining tumor extent, assessing involvement of surrounding tissues, and identifying features suggestive of malignant transformation.[5] In our patient, MRI demonstrated a well-defined soft tissue mass without bone involvement or radiological evidence of malignancy, facilitating surgical planning.

Although targeted therapies such as selumetinib have demonstrated promising results in reducing the volume of inoperable plexiform neurofibromas, surgery remains the treatment of choice for accessible symptomatic lesions causing functional impairment.[6,7,10] Complete excision can provide significant symptomatic relief, improve function, and reduce the risk of local recurrence. However, surgical management may be challenging because of tumor vascularity, infiltration of surrounding tissues, and proximity to major neurovascular structures.[10]

Histopathological examination remains the gold standard for diagnosis and is essential to exclude malignant peripheral nerve sheath tumors, which develop in approximately 8-13% of patients with NF1 during their lifetime.[10] In the present case, histopathology confirmed a benign neurofibroma without atypia or malignant features. Complete surgical excision resulted in excellent functional recovery and satisfactory cosmetic outcomes without postoperative complications or recurrence during follow-up.

CONCLUSION

Giant neurofibromas around the knee are rare manifestations of Neurofibromatosis Type 1 and can lead to significant functional impairment. Thorough clinical evaluation, appropriate imaging, and complete surgical excision are crucial for optimal management. Early intervention can improve mobility, quality of life, and long-term outcomes.

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