Journal of Pakistan Association of Dermatologists. 2024;**34(4 Suppl):**S134-S137.

Case Report

Neurofibromatosis type 1 presenting with tender enlarging plexiform neurofibroma in the lumbar region of an adolescent

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Abstract Plexiform neurofibromas are soft tissue tumours that arise from peripheral nerves and are often seen in patients with Neurofibromatosis type 1 (NF1). We report a case of a 15-year-old girl with NF1 who presented with a painful, enlarging swelling extending laterally from the right lumbar region to the anterior chest wall. Multidisciplinary approach was undertaken; neurosurgeons and plastic surgeons were involved. This case emphasizes that early diagnosis and timely intervention can alter the long-term outcome in these patients.

Key words
Plexiform neurofibroma; Neurofibromatosis Type 1; Lumbar region; Malignant transformation; Adolescent.

Introduction

Neurofibromatosis type 1 (NF1) is a genetic disorder characterized by the development of multiple benign tumors, particularly neurofibromas.[1] Plexiform neurofibromas are benign tumors commonly associated with Neurofibromatosis Type 1 (NF1). Although benign, these tumors can undergo malignant transformation into malignant peripheral nerve sheath tumors (MPNSTs), a rare but aggressive form of sarcoma. Early recognition of malignant transformation is crucial for improving outcomes.[2] These tumours can cause significant morbidity due to their size and potential for malignant transformation[3]. This report chronicles the clinical course and management of a lumbar plexiform neurofibroma in an adolescent girl, emphasizing the importance of longitudinal monitoring.

Manuscript Received on: October 11, 2024
Accepted on: December 11, 2024
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Case report

A 15-year-old female diagnosed with Neurofibromatosis type 1 based on the presence of multiple café-au-lait spots (5cm to 15cm in size), axillary freckling, lisch nodules, cutaneous plexiform neurofibromas and a positive family history presented with a three-years history of progressive swelling in the right lumbar region following an injury. Initially painless, the swelling became increasingly painful over the past one year with tenderness and difficulty in lying on the back. There were no constitutional symptoms of fever or weight loss, but she reported significant discomfort with movement. On examination, the patient appeared well but in mild distress due to pain. A firm, irregularly shaped mass, approximately 12 cm in greatest diameter, was palpated extending from the left lumbar region to the anterior chest wall. The mass was tender on palpation and had a firm, fixed consistency with ill-defined margins. Overlying skin was dry and hyperpigmented

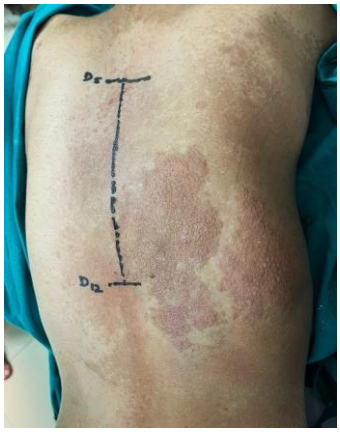

Figure 1 Ill-defined, subcutaneous mass involving right lumbar area along with hyperpigmentation of skin of back.

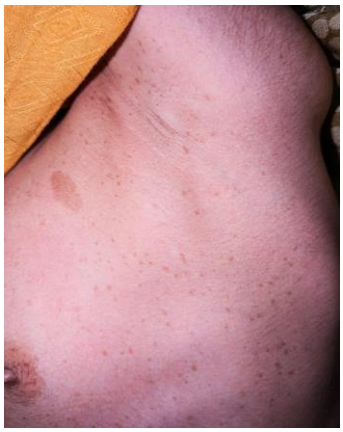

Figure 2 Left axillary freckling and café au lait macules on trunk.

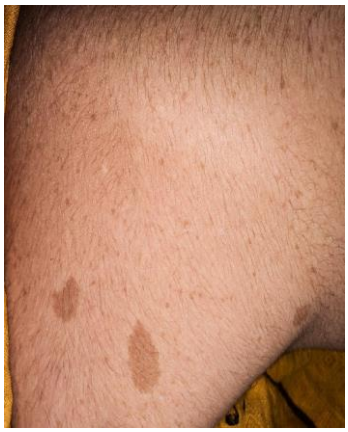

Figure 3 Multiple freckles and café au lait macules on posterior aspect of left upper arm.

(Figure 1). Multiple café-au-lait spots were present on the body along with axillary freckling **(Figures 2,3)**. There were no signs of respiratory compromise, blurring of vision, bone deformities or neurological deficit.

Computed Tomography (CT) scan chest with contrast showed soft tissue density areas causing widening of right neural foramina associated with minimally enhancing subcutaneous nodules at the level of D7 and D8 vertebral bodies **(Figure 4)**, suggestive of neurofibromatosis.

Magnetic Resonance Imaging (MRI) dorsal spine showed a large, infiltrative mass with multiple elongated contiguous altered signal intensity areas in para-spinal region more marked in the right lumbar region, extending along posterolateral chest wall opposite D5 to D9 levels, with no post contrast enhancement **(Figure 5)**, suggestive of neurofibromatosis.

A multidisciplinary team was convened. The patient underwent extensive surgical resection of the plexiform neurofibroma with primary closure of surrounding tissues **(Figure 6)**. Biopsy was taken for histopathology. Histopathological analysis showed bundles of benign looking cells with wavy and elongated comma shaped nuclei, in a storiform pattern **(Figures 7,8)**.

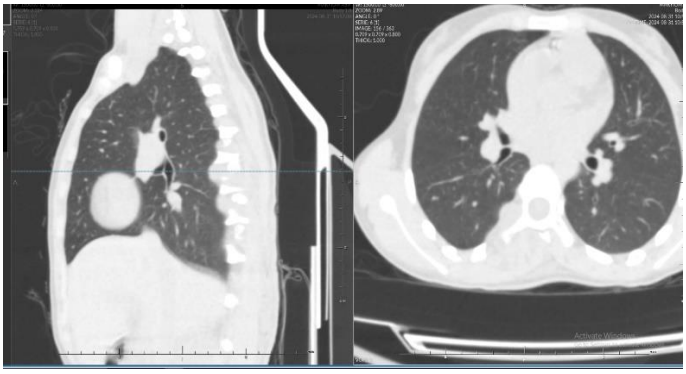

Figure 4 CT scan chest with IV contrast showing widening of right D7 neural foramen due to soft tissue nodule (most likely neurofibroma).

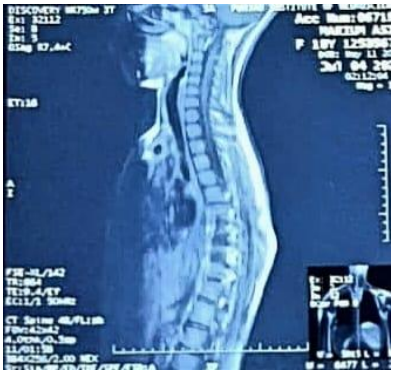

Figure 5 MRI dorsal spine showing multiple elongated contiguous altered signal intensity areas in para-spinal region more marked in the right lumbar region, extending along posterolateral chest wall opposite D5 to D9 levels.

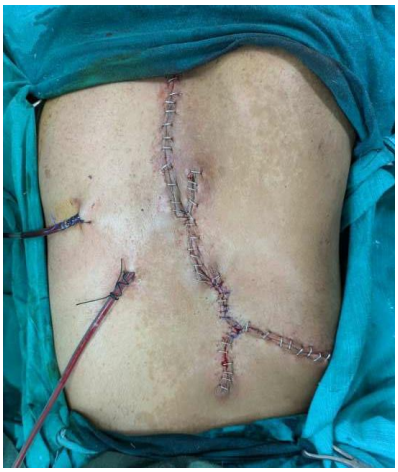

Figure 6 Post-resection closure with drains.

There was subtle myxoid change in the background. No atypia, necrosis or mitosis was seen. No evidence of malignant transformation was seen. Based on these findings, a diagnosis of plexiform neurofibroma was made.

Discussion

This case underscores the necessity for continuous monitoring of plexiform neurofibromas, particularly in adolescents with NF1. The transition from benign growth to malignant transformation necessitates prompt intervention, as illustrated by the patient's clinical course. The incidence of malignant peripheral nerve sheath tumours (MPNSTs) arising from neurofibromatosis type 1 (NF1) is estimated to be between 8% and 13%.[3] Patients with NF1 are at a significantly higher risk of developing these aggressive tumours compared to the general population. Furthermore, the prognosis for NF1 patients with MPNSTs is generally poorer than for those without NF1 due to the aggressive nature of the tumours.[4] The pathogenesis of this transformation is believed to be related to mutations in the NF1 gene, which leads to the loss of neurofibromin, a protein that normally regulates cell proliferation.[5] Without neurofibromin, uncontrolled growth can result, leading to tumour development and, in some cases, malignant transformation.[6]

Clinically, the challenge lies in differentiating between benign neurofibromas and early-stage MPNSTs. Indicators such as rapid growth, new-onset pain, or changes in consistency should raise suspicion for malignancy. Unfortunately, MPNSTs are highly aggressive, and their prognosis is often poor, especially when diagnosis is delayed. The current standard treatment is wide surgical excision, often combined with radiation and chemotherapy, although outcomes remain suboptimal due to frequent recurrence and metastatic potential[7]. Given these challenges, regular monitoring of NF1 patients is critical. Advanced imaging techniques, particularly MRI, are invaluable in detecting changes in tumour size or characteristics that may indicate malignant transformation.[8] Furthermore, emerging therapies, such as targeted inhibitors of the RAS/MAPK pathway and immunotherapies, are being investigated and hold promise for improving the outcomes of NF1-associated MPNSTs.[7]

Conclusion

Plexiform neurofibromas are an important part of Neurofibromatosis. Their longitudinal

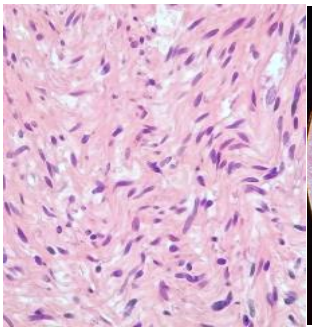

Figure 7 Histopathology (H&E at 4x) showing well circumscribed, non-encapsulated, growth with cells in storiform pattern.

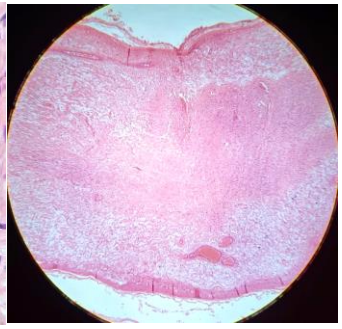

Figure 8 Histopathology 40x showing bundles of elongated cells with com shaped nuclei.

assessment is critical for early identification of malignancy. Clinicians should remain vigilant for changes in size or symptoms, as timely surgical and medical management can lead to improved patient outcomes.

Acknowledgement We are grateful to patient's family for their cooperation and compliance to investigations and management. Patient's parents gave consent for sharing her data and photos.

Declaration of patient consent The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship None.

Conflict of interest Authors declared no conflict of interest.

Authors' contribution

RM,HT,ANC,FA: Diagnosis and management of the case, critical review of the manuscript, has given final approval of the version of the manuscript to be published.

JBT,SB: Identification and management of the case, manuscript writing, has given final approval of the version of the manuscript to be published.

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