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Child Plexiform Neurofibroma of the Lumbar Region: Case Report

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Abstract **Case Report**

Neurofibromas are benign tumors that develop from the nerve roots and plexuses of the spinal nerves. Plexiform neurofibromas, which are pathognomonic of Neurofibromatosis Type 1 (NF1), are typically slow-growing and can present with a variety of symptoms depending on their location. This article reports the case of a 7-year-old child with a history of scoliosis who presented with a swelling in the lumbar region. Clinical examination revealed painless subcutaneous nodules, multiple café au lait spots, and hypertrichosis. Ultrasound showed hypo-echoic, well-limited, hypervascular nodules, while thoracolumbar MRI revealed T2 hyperintense nodular thickening of the dorsolumbar roots and paraspinal soft tissues, consistent with plexiform neurofibromas. These findings confirmed the diagnosis of NF1 with plexiform neurofibromas. The discussion highlights the importance of clinical examination and advanced imaging, particularly MRI, in diagnosing NF1 and assessing the extent of the lesions. Management primarily involves symptom monitoring, as complete surgical resection is often challenging. Early diagnosis and long-term follow-up are critical to managing complications such as tumor progression, skeletal abnormalities, and malignant transformation.

Keywords: Neurofibromatosis Type 1, Plexiform Neurofibromas, Spinal Nerves.

INTRODUCTION

Neurofibromas are benign tumors that develop from the roots and plexuses of the spinal nerves. They can be unilateral or bilateral, occasionally multifocal, superficial, or deep.

Neurofibromas can be either cutaneous or plexiform. Plexiform neurofibromas are pathognomonic of NF1. They are typically slow-growing tumors. Their symptomatology depends on their location. We report the case of a 7-year-old child presenting with a swelling in the lumbar region.

CASE REPORT

This is a 7-year-old patient, followed for scoliosis, who consulted for a swelling of the lumbar region. Clinical examination revealed painless subcutaneous nodules with no inflammatory signs. Skin examination revealed multiple ‘café au lait’ spots on the back and limbs (more than 5) associated to hypertrichosis (Fig-1).

A soft tissue ultrasound was ordered and showed subcutaneous nodular formations, oval in shape, well limited in the lumbar region, hypo-echogenic, heterogeneous and hyper-vascularized on color Doppler (Fig-2).

A thoracolumbar MRI was performed to characterize the lesions described above. It revealed a scoliotic attitude with stepped nodular thickening of the nerve roots from D6 to L1 in T2 hypersignal (Fig-3) with contrast after injection of Gadolinium; a nodular thickening of the intercostal spaces and of the left paraspinal soft tissues in T2 hypersignal (Fig-4) was also observed.

In view of this clinical picture and the imaging appearance, the diagnosis of neurofibromatosis type I was evoked with plexiform neurofibromas of the dorsolumbar roots and neurofibromas of the subcutaneous soft tissues.

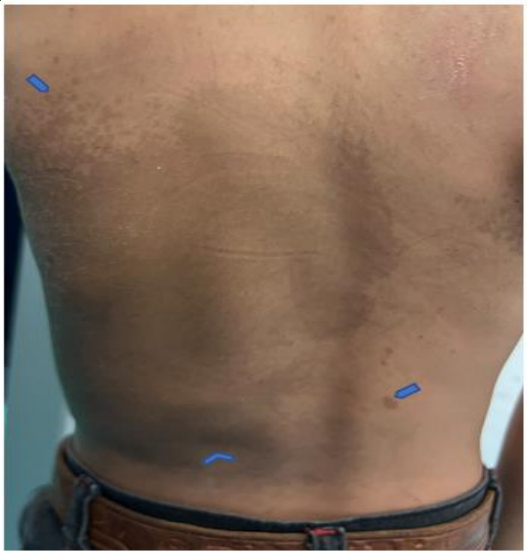

Fig-1: Capture of the child's back illustrating: a left lumbar subcutaneous swelling with hypertrichosis and hyperpigmentation without telangiectasia or vascular-nervous disorders and multiple 'café-au-lait' spots larger than 5 mm

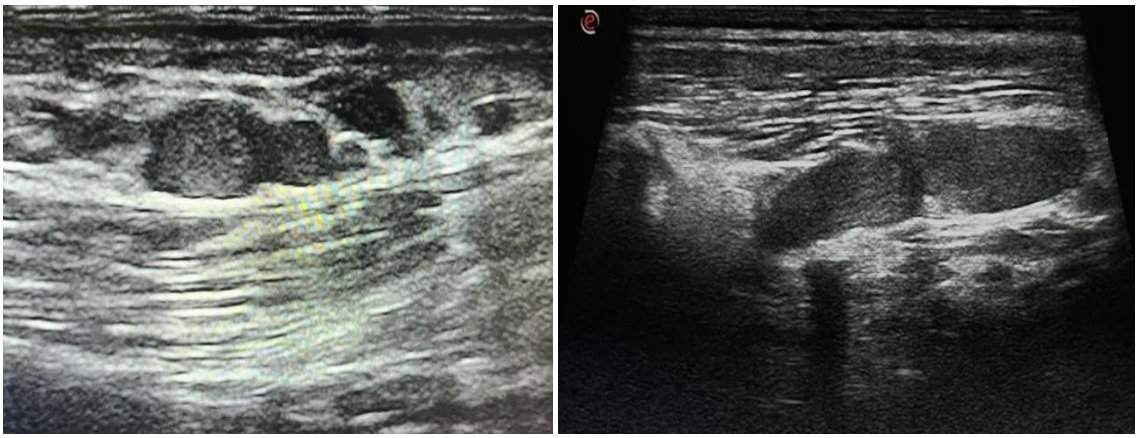

Fig-2: Soft tissue ultrasound showing subcutaneous nodular formations, well limited in the lumbar region related to subcutaneous neurofibromas

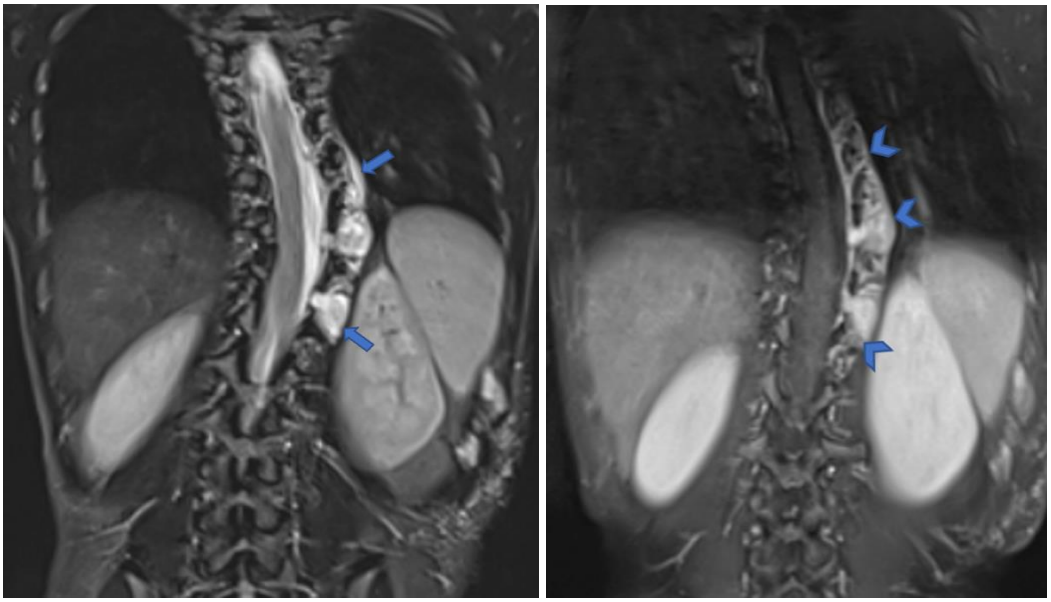

Fig-3: Scoliotic attitude with nodular thickening staged from D6 to L1 appearing hyperintense on T2 (arrows) enhanced after contrast injection (arrow heads)

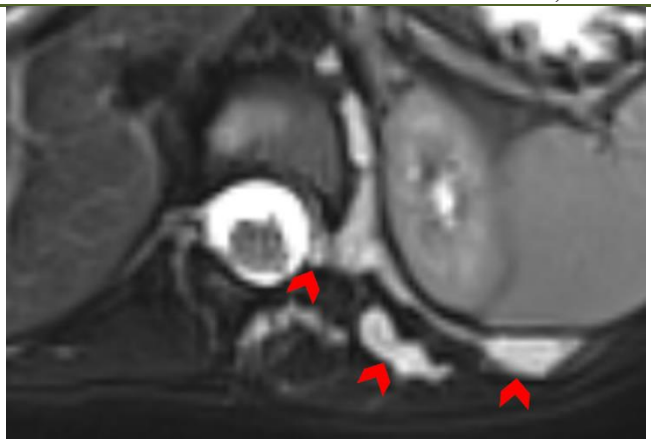

Fig-4: Root nodular thickening of intercostal spaces and paravertebral soft tissue in hypersignal T2

DISCUSSION

Neurofibromas are benign peripheral nerve sheath tumors originating from Schwann cells, fibroblasts, and other neural crest-derived cells. They are often associated with Neurofibromatosis Type I (NF1), an autosomal dominant genetic disorder caused by mutations in the NF1 gene located on chromosome 17. Plexiform neurofibromas, a specific subtype, are pathognomonic of NF1 and are characterized by diffuse and infiltrative growth along nerve bundles and plexuses. They can involve multiple tissue planes, including subcutaneous tissues, nerves, and deep paraspinal regions, as observed in this case [1, 2].

Clinical Features and Diagnostic Challenges:

Plexiform neurofibromas often manifest in childhood and are typically slow-growing. Clinical features depend on the tumor's location and size. In our case, the child presented with a swelling in the lumbar region, painless subcutaneous nodules, and hallmark NF1 features such as multiple café au lait spots and hypertrichosis. These findings align with diagnostic criteria for NF1, which include the presence of multiple café au lait spots, neurofibromas, or distinctive skeletal abnormalities like scoliosis, also seen in this patient [3].

Imaging in Diagnosis:

MRI is the imaging modality of choice for characterizing plexiform neurofibromas due to its superior soft tissue contrast. The thoracolumbar MRI in this case revealed the characteristic T2 hyperintense nodular thickening of nerve roots, which is highly suggestive of plexiform neurofibromas [4]. Additionally, the identification of contrast enhancement with gadolinium provided further confirmation of their neural origin. Ultrasound, although less sensitive, supported the diagnosis by revealing hypo-echoic, well-defined, and hypervascular nodules in the subcutaneous tissues [5]. Together, these imaging modalities provide critical information on the extent of the lesions and guide management.

Complications and Prognosis

While plexiform neurofibromas are benign, they carry a risk of malignant transformation into malignant peripheral nerve sheath tumors (MPNST), reported in approximately 10% of cases [6]. Other complications include compression of adjacent structures, neurological deficits, and significant disfigurement. Early identification and regular monitoring of these lesions are crucial to managing these risks. In this case, the involvement of the dorsolumbar roots and paraspinal tissues raises the possibility of progressive scoliosis and potential spinal cord compression, emphasizing the need for multidisciplinary care.

Management Considerations:

Management of plexiform neurofibromas primarily involves symptomatic treatment and monitoring, as complete surgical resection is often challenging due to their infiltrative nature and proximity to critical structures. Novel therapies, including MEK inhibitors such as selumetinib, have shown promise in reducing the size and symptomatic burden of plexiform neurofibromas in pediatric patients [7]. Long-term follow-up is essential to monitor for complications, including tumor progression, skeletal abnormalities, and potential malignant transformation.

CONCLUSION

This case highlights the classic presentation of NF1 with plexiform neurofibromas, underscoring the importance of clinical examination and advanced imaging in establishing the diagnosis. Early diagnosis and close follow-up are essential for optimizing outcomes and minimizing complications in children with NF1.

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