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Case Report

Pathology Section

Retroperitoneal Malignant Peripheral Nerve Sheath Tumour: A Rare Case Report

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ABSTRACT

Malignant nerve sheath tumours (MPNST) are rare neoplasias and retroperitoneal cases are fairly rare and clinically difficult to be detected, but they are very agressive neoplasias. MPNST are frequently seen in head, neck and upper extremities. In patients with NF1; MPNST, a poor-prognostic lesion, may result from a malignant degeneration of a former plexiform neurofibroma. It is necessary to be aware of a potential malignancy in patients diagnosed with plexiform neurofibroma.

We present a 21-year-old female with a diagnosis of MPNST. The patient was admited to the hospital because of a tumour in the subcutaneous region on her left buttock. The surgeon's clinical diagnosis was lipoma. After the pathological examination of biopsy specimen, the lesion was identified as "plexiform neurofibroma" and then the patient was diagnosed with Neurofibromatosis Type 1 (NF1). Simultaneously, another mass on the retroperitoneal region was identified as malignant peripheral nerve sheath tumour (MPNST).

Keywords: Neurofibromatosis Type 1(NF1), Retroperitoneal mass, Plexiform neurofibroma

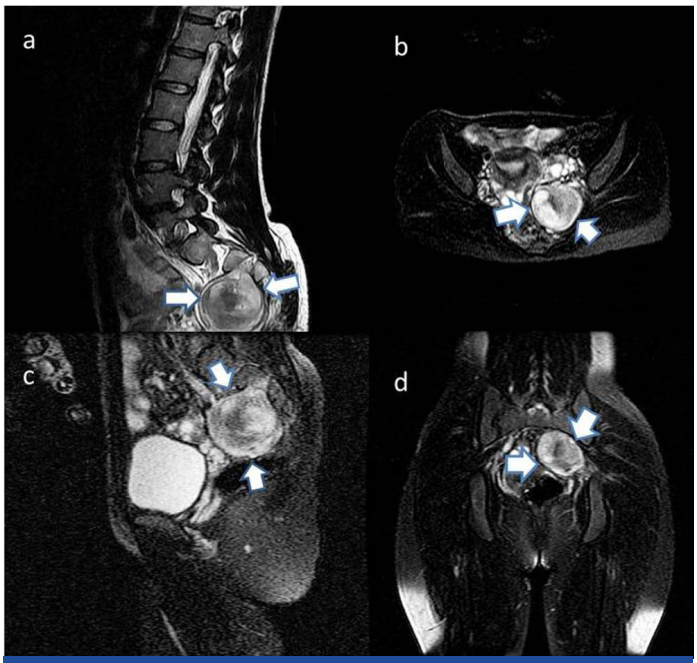
CASE REPORT

A 21-year-old female patient with left hip pain, ongoing for 5-6 months and painful swelling in the left buttock, was admitted to hospital. No other symptoms, including muscle atrophy, weakness and functional limitation were noted. On examination, a partially mobile subcutaneous mass was observed in the left hip. An informed consent was taken for the surgical procedure and the patient was operated immediately. The mass was removed in toto, under local anaesthesia. On macroscopic examination, a 2x1,5x1,5cm non-encapsulated mass was noted. On microscopic examination, a non-encapsulated lesion from the reticular dermis extending into the subcutaneous fat tissue was observed. Enlarged nerve branches formed tortuous masses within delicate musinous stroma in the lesion. These nerve branches involved long cells with wavy dark nucleus. The cells were intertwined. In these cells, changes such as nuclear atypia and an increased number of mitotic activity that may be associated with malignancy, were not observed.

On the immunohistochemical examination, the cells within the nerve fascicles were positive for S100. Based on these findings, the lesion was diagnosed as Plexiform Neurofibroma. Plexiform Neurofibromas are common lesions in NF1. For this reason, the patient was recalled for a complete clinical examination. It was noted that the patient's father had NF1. On physical examination, two cafe-au-lait macules on the forearm, approx 20mm in diameter, were noted. Lisch nodules in both eyes were noted. National Institute of Health diagnostic criteria for NF1 (1997) was used and the patient was diagnosed with NF1. It was noted that the patient had been suffering from left-hip pain even after excision of the subcutaneous mass. Thus an MRI of the lumbar and lower abdomen revealed a second mass in the left half of the pelvis-in the left-midline pushing the left ovary to the inferior.

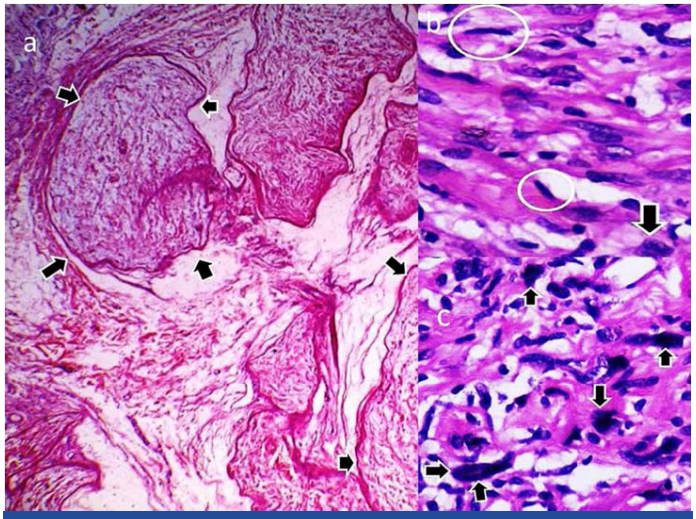
In lumbar spinal MRI, heterogeneous presacral mass was seen in the sagittal sections. In pelvic MRI, presacral partially enhancing mass was seen respectively in transverse, sagittal and coronal sections. The mass extend till S2 neural foramen [Table/Fig-1a-d].

A surgery was planned and it was found that the mass originated in sacral roots. The mass was totally excised. It measured 6x5x4.5cm,

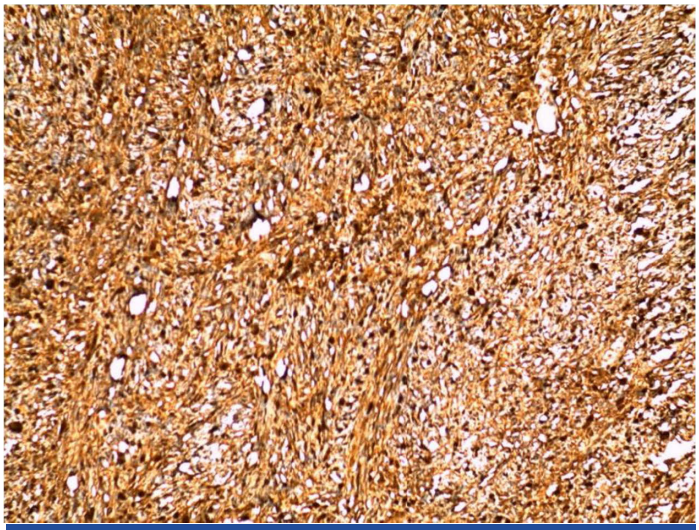


[Table/Fig-1a-d]: a) Sagittal LS MRI; b) Transverse LS MRI; c) Sagittal LS MRI; d) Coronal Pelvic MRI

had a pseudocapsule, was white-grey in colour with mucoid appearance and had focal areas of haemorrhage and necrosis. On microscopy, it consisted of spindle-shaped cells and forming fascicles and -rarely- whorled structures. In some areas, the nuclei were observed to be palisading. Nucleus were seen to be wavy and oblique. Pleomorphic tumour cells with rare relatively uniform tumour cells and with wavy nucleus were seen. However, hyperchromatic nuclei and marked pleomorphism were seen most of the tumour cells in the section. Also, most of the tumour cells revealed increased mitosis. Around the atypical tumour cells coagulation necrosis was noted. In the immunohistochemical study of tumour cells with S100 and vimentin positive staining was observed. Desmin and SMA were negative [Table/Fig-2-4]. Ki67 proliferative index was around 20-25%. On the basis of the histopathological



[Table/Fig-2]: a) Plexiformneuro fibroma, enlarged nerve fascicles (H&E 4X); b) MPNST, pleomorfic tumour cells (arrow), rare relatively uniform tumour cells with wavy nucleus (ring) (H&E 40X); c) MPNST, hyperchromatic nuclei and cellular pleomorphism (H&E 40X)



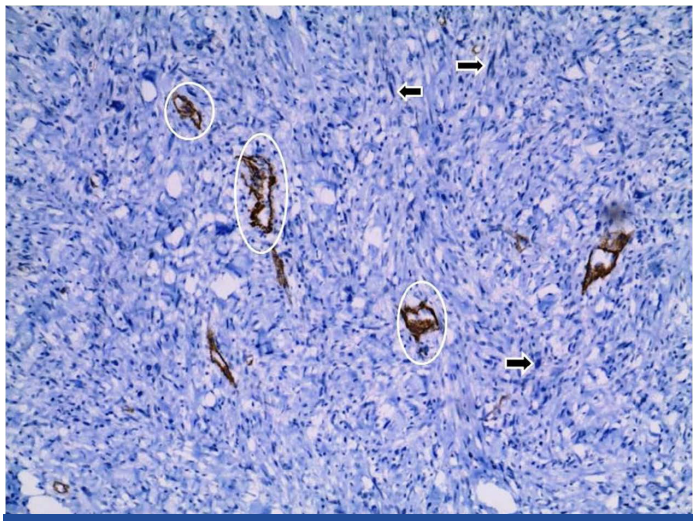
[Table/Fig-3]: MPNST, Cytoplasmic and nuclear immunostaining for S100X4

and immunohistochemical features, a diagnosis of MPNST was made. The patient has been followed up clinically and radiologicaly and has been recurrence free for a year.

DISCUSSION

NF1 is an autosomal dominant disorder [1-3]. The frequency of benign and malign tumour incidence is increased in NF1 [3]. Neurofibromas are the most common benign tumours in NF1 [1]. Besides, MPNSTs are seen in NF1 [3-6]. Unlike other neurofibroma types, plexiform neurofibroma carries increased risk for MPNST [1,4]. MPNSTs are quite rare neoplasms which cover 10-12% of all soft tissue sarcomas. While the rate of MPNST is 0.001% in the general population, with NF1 patients the rate is approximately 2-5% [3,7,8]. MPNST is frequently seen in people of 2nd and 3rd decade. In NF1cases, MPNST is seen at a younger age than in the general population [3]. Frequently asymptomatic mass is seen in MPNST. Growing mass and pain are the most important clinical symptoms. MPNSTs are seen in head, neck and upper extremities. Only %1 of MPNSTs is retroperitoneal [9].

Retroperitoneal mass is asymptomatic in the early stages [10]. Retroperitoneal location is clinically difficult to be detected [9]. ZH Meng et al., presented a retroperitoneally located MPNST which was detected by USG as a 'hepatic neoplasm'. Later FNAC revealed presence of spindle shaped cells and a few gangliocytes without any evidence of malignancy [9]. Dakshyani S Mirhale et al., published a retroperitoneal case of MPNST, appearing as a palpable lump in the



[Table/Fig-4]: MPNST, negative staining for SMA in the tumour cells (arrow) and positive reaction in the vessel wall (ring) X10

left upper abdomen. On exploratory laparotomy, it was found that left kidney was involved as well as the spleen. Hence with the mass excision, splenectomy and left nephrectomy was performed [10]. In our case, the mass was near the left ovary and deeply located. Despite this, an early diagnosis was possible before spreading to the surrounding tissues.

Particularly with deeply located lesions like retroperitoneal location, radiological tests, such as CT and MRI, are helpful in diagnosis [7]. In our patient, retroperitoneal located mass was imaged with MRI. MPNSTs have no specific laboratory findings or morphological features, making the preoperative diagnosis difficult.

In most cases, the diagnosis depends on the pathologic and immunohistochemical studies [9,11]. However, it is yet difficult to conclude a diagnosis of MPNST in the light of only microscopic findings [7]. If the histomorphologic findings in a malign soft tissue tumour consisting of spindle cells make one thinks MPNST, there should be at least one of the two following criterions in order to conclude a diagnosis of MPNST: 1) Tumour should arise in a patient with NF1; 2) Tumour should appear to arise in a nerve or should develop in a previous neurofibroma [7].

MPNST may develop de novo or it may develop as a result of malignant transformation of previous plexiform neurofibroma. We cannot be sure of it but, it is prudent that one should always search for the existence of malignant masses in patients diagnosed with plexiform neurofibroma. MPNSTs have a high malignant potential, recurrence and distant metastasis are common [3,9]. Treatment is predominantly surgical. Complete surgical excision of the tumour should be made with negative margins [3]. In our patient, the mass was excised with negative surgical margins.

CONCLUSION

In patients with NF1; MPNST may result from a malignant degeneration of a former plexiform neurofibroma. It is necessary to be aware of a potential malignancy in patients diagnosed with plexiform neurofibroma.

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