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Case Report

Intraosseous malignant peripheral nerve sheath tumor of the sacrum in a patient with neurofibromatosis type I

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ABSTRACT

Malignant intraosseous peripheral nerve sheath tumor is a very rare malignancy most commonly seen in patients with neurofibromatosis type 1. This tumor almost exclusively occurs in the maxillofacial region, with manifestation of this tumor in other regions of the skeleton infrequently reported. We describe a 23-year-old female with previously undiagnosed neurofibromatosis type 1 presenting with lower extremity weakness, paresthesias, and bowel/bladder symptoms. The patient had an aneurysmal lytic bone lesion centered in the upper sacrum with invasion of the L5 vertebral body. On MRI, the lesion was homogeneously isointense to muscle on T1, heterogeneously hyperintense to muscle on T2, and demonstrated homogeneously avid contrast enhancement. Multiple additional small lesions with similar imaging characteristics were identified in the paraspinal soft tissues. Low grade malignant peripheral nerve sheath tumor of the sacrum was diagnosed on biopsy. The patient was treated with sacral resection and radiation therapy for local disease control.

Introduction

Malignant peripheral nerve sheath tumor (MPNST) is a rare mesenchymal tumor comprising only ~2% of all sarcomas [1]. MPNST is most commonly associated with patients with a history of neurofibromatosis type 1 and typically arises from a pre-existing plexiform neurofibroma [2]. Most MPNSTs occur in the soft tissues with intraosseous origin of MPNST only sporadically reported in the literature [3]. When intraosseous MPNST does occur, it is most frequently reported in the head and neck, although, other locations like the metacarpals, calcaneus, and phalanges, have been reported [4–6]. We report a case of large MPNST arising from the sacrum in a patient

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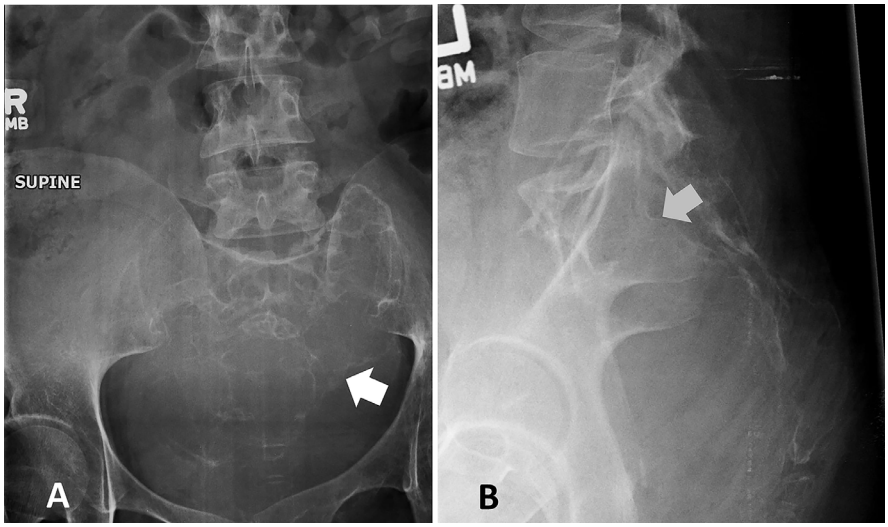

Fig. 1 – AP (A) and Lateral (B) radiographs of the lower lumbar spine and sacrum. Severe demineralization and cortical destruction is present throughout the upper sacrum (white arrow). Demineralization and cortical disruption consistent with tumor invasion can be seen along the inferior and posterior aspects of the L5 vertebral body on the lateral radiograph (grey arrow).

with previously undiagnosed neurofibromatosis type 1. Our case demonstrates MPNST should be considered in the setting of a large sacral mass and neurofibromatosis.

Case report

A 23-year-old female presents with severe lower back pain after fall from standing 2 weeks prior. The patient reported 2-year history of vague lower back pain with increasing difficulty walking over the preceding few months due to developing lower extremity paresthesias and weakness. Since her fall, the patient had developed intermittent fecal incontinence as well as increased urinary frequency and urgency. Otherwise, the patient reported no prior medical conditions. There was no family history of neurofibromatosis. Physical exam was remarkable for 3 out of 5 strength in the right quadriceps/iliopsoas muscles and 4 out of 5 strength of the left quadriceps/iliopsoas muscles. The patient was also found to have multiple café-au-lait patches on the trunk and extremities as well as multiple small, mobile subcutaneous nodules initially believed to be subcutaneous lipomas. No cutaneous nodules, iris hamartomas, or visual disturbances were noted on physical exam.

Radiographs of the lumbar spine were obtained for initial evaluation, which demonstrated extensive destruction of the sacrum and L5 vertebral body (Fig. 1). CT of the pelvis without contrast (Fig. 2) and MRI of the sacrum with and without contrast (Fig. 3) were then performed to further evaluate the lesion. CT demonstrates aneurysmal expansion of the upper sacrum by purely lytic soft tissue mass containing a few thin calcified septations. The mass directly invaded the L5 vertebral body and cause severe narrowing of the spinal canal. The mass was homogeneously isointense to skeletal muscle

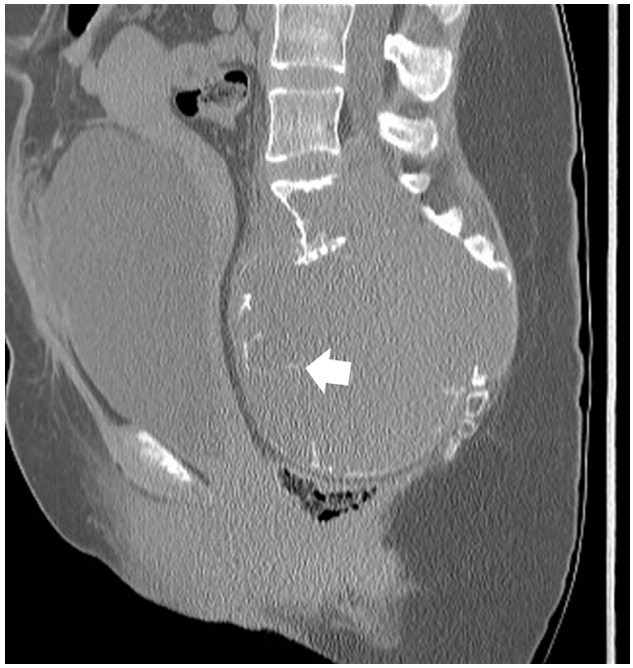

Fig. 2 – Sagittal noncontrast CT of pelvis at midline demonstrates a large mass centered within the sacrum. The mass causes aneurysmal expansion of the sacrum with severe cortical thinning and contains a few thin, septal-like calcifications (arrow).

on T1 images and heterogeneously hyperintense to skeletal muscle on T2 images with areas of fluid-like T2 signal. The mass demonstrated near uniform avid enhancement on post-contrast imaging. Multiple additional small soft tissue masses with similar signal characteristics as the primary sacral mass

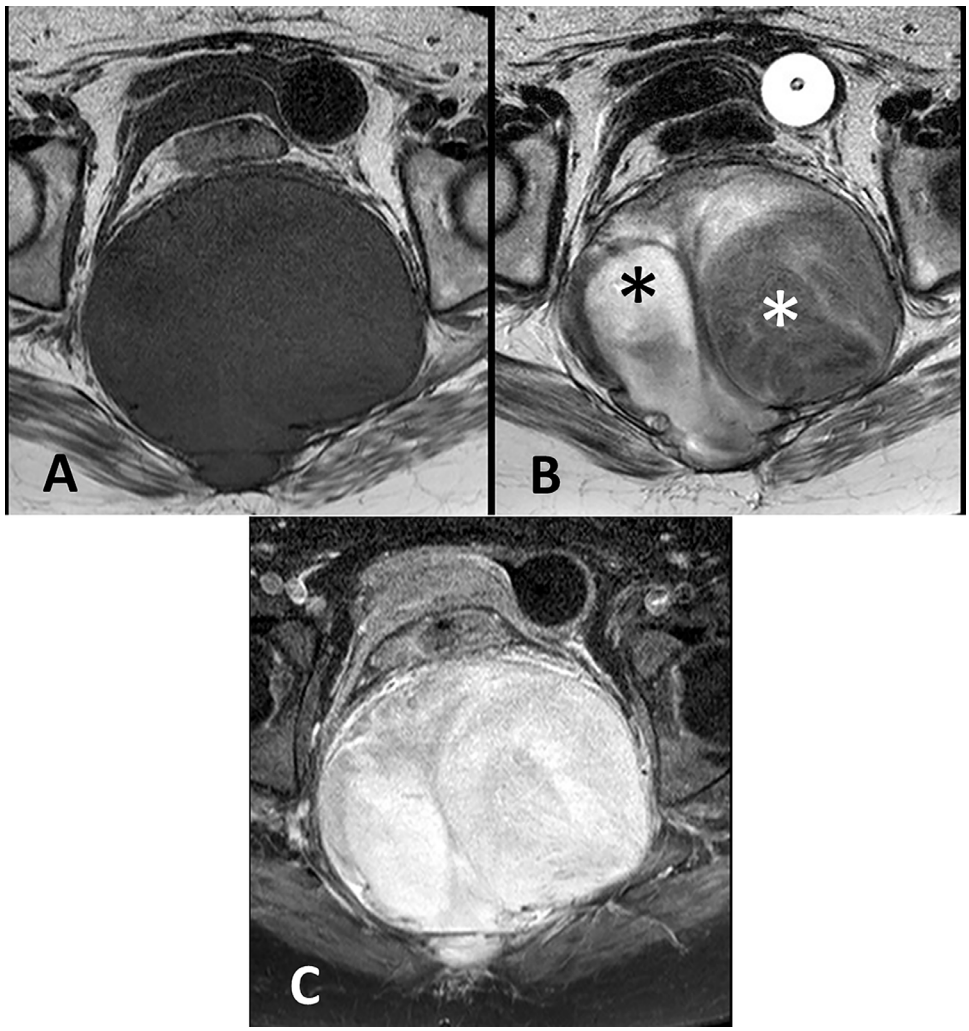

Fig. 3 – Axial T1 (A), T2 (B), and T1 postcontrast fat suppressed (C) sequences of the sacrum at the S3-S4 level. The lesion is homogeneously isointense to skeletal muscle on T1 and heterogeneous on T2 with areas of fluid-like signal (black asterisks) and areas that are slightly hyperintense to skeletal muscle (white asterisks). The lesion demonstrates slightly heterogeneous diffuse avid enhancement.

were identified in the paraspinal soft tissues of the chest, abdomen, and pelvis (Fig. 4).

Core needle biopsy of the lesion was performed, which demonstrated a spindle cell neoplasm with up to 8 mitoses per 10 high-power fields (Fig. 5A). The lesion had positive staining for beta-catenin, S100, and CD34 as well as negative staining for pan-keratin, CD117, SMA, HMB45, SOX-10, and neurofilament. The ki-67 positive staining ranged from <5% to 30% (Fig. 5B). Histology findings were consistent with a low-grade malignant peripheral nerve sheath tumor. The patient subsequently underwent sacral resection with incomplete resection of the sacral mass, posterior lumbopelvic fusion, and fibular allograft placement (Fig. 6). A staging PET/CT scan did not show any evidence of metastatic disease. The patient was subsequently scheduled to receive radiation therapy for local control of residual tumor. Based on the presence of multiple café-au-lait spots, malignant peripheral nerve sheath tumor, and multiple soft tissue masses consistent with neurofibromas on MRI, the patient was diagnosed with neurofibromatosis type 1.

Discussion

Malignant peripheral nerve sheath tumor is a rare soft tissue sarcoma arising from a peripheral nerve, typically in patients between the age of 20 and 50 [3,7]. There is a strong association between MPNSTs and neurofibromatosis. Approximately 25%-50% of cases are associated with NF-1, and NF-1 patients carry a lifetime risk of approximately 8%-12% for developing MPNSTs [8,9]. The hallmark feature of NF-1 is the neurofibroma, of which there are 3 main types: cutaneous, subcutaneous, and plexiform [10]. Of these 3 types of neurofibromas, MPNST's arise most frequently from plexiform neurofibromas [8,11]. MPNSTs not originating from a neurofibroma can

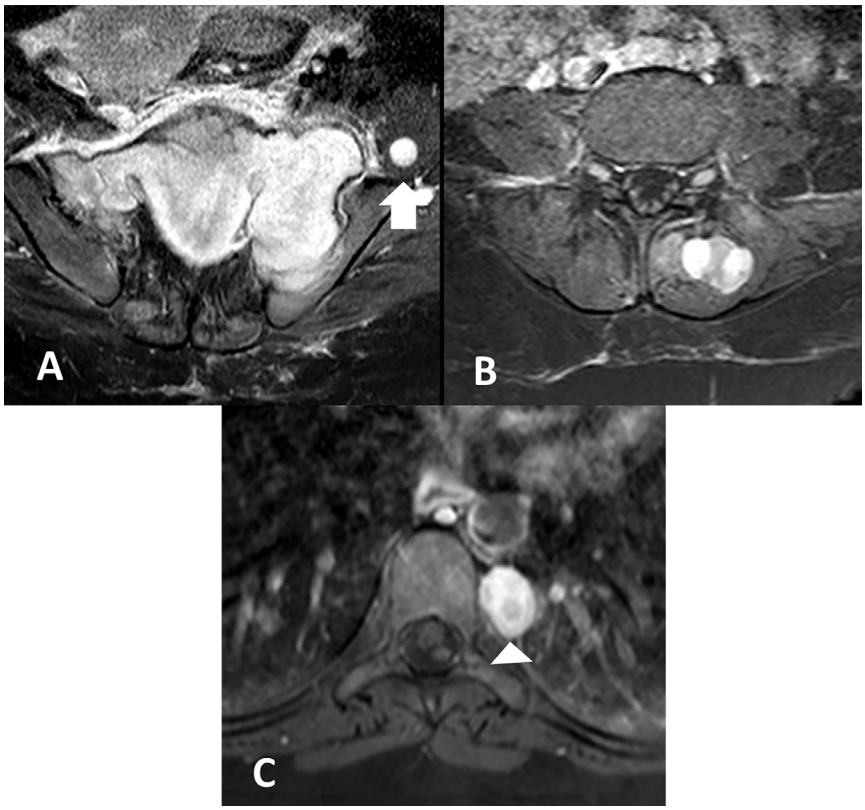

Fig. 4 – Axial T1 postcontrast fat suppressed images at the level of the S1 (A), L3 (B), and T10 (C) vertebrae. Avidly enhancing soft tissue masses are present in the left psoas (white arrow), left lumbar paraspinal muscles (grey arrows), and left posterior mediastinum (arrowhead).

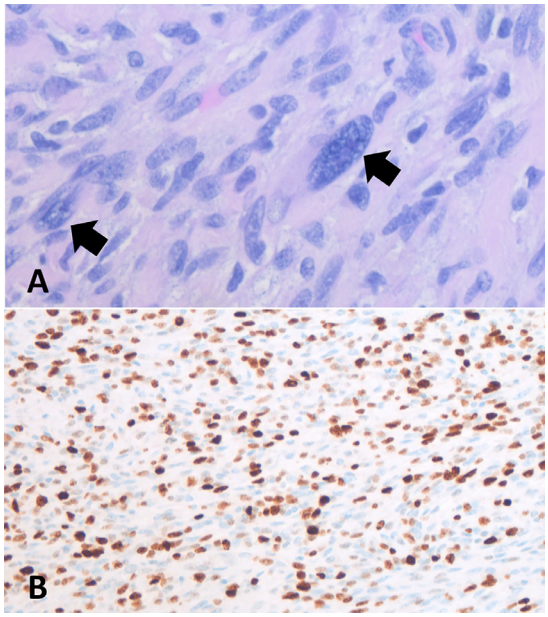

Fig. 5 – (A) High degree of cellularity and nuclear atypia (arrows) on hematoxylin and eosin stain (400× magnification). (B) Immunohistochemical stain with antibodies against Ki67 shows a large number of brown positive nuclei, denoting a high proliferation rate (100× magnification).

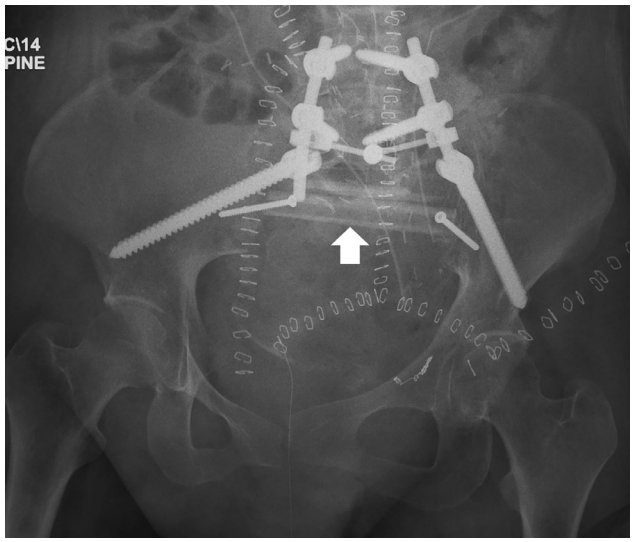

Fig. 6 – Postoperative AP radiograph of the pelvis after sacral resection with lumbopelvic fusion. Fibular allograft spans the iliac bones (arrow).

arise de novo from normal peripheral nerves or can occur secondary to prior radiation therapy [3,7].

Intraosseous peripheral nerve sheath tumors (PNSTs) are rare. The tumor may be a primary intraosseous PNST

arising from myelinated intraosseous nerve fibers or congenital rests of neural crest tissue within the bone, or the tumor may be a secondary intraosseous PNST from erosion into the bone on a PNST within a neuroforamin [3]. A majority of cases of intraosseous PNST arise from the head and neck [3,12]. This is likely due to the large number of intraosseous nerves located in neuroforamina in the head and neck [3].

Differentiation of malignant from benign PNST can be challenging. Clinical findings suggesting malignant degeneration include pain and discomfort at the tumor site in addition to new or worsening neurologic symptoms [8]. In a study by Wasa et al, 4 MRI findings were found to be helpful in distinguishing MPNST from benign neurofibromas: large size of the mass, a peripheral enhancement pattern, perilesional edema, and intratumoral cystic change [13]. The presence of 2 of these 4 MRI findings was found to have a specificity of 90% and sensitivity of 61% for identifying MPNST. Heterogeneity on T1 imaging was also found to be significantly more common in MPSNTs than neurofibroma, although, this finding was not included as one of the 4 distinguishing criteria [13]. Diffuse-weighted imaging has also been shown to be useful in differentiating between benign and malignant peripheral nerve sheath tumors with benign PNSTs typically having a high ($>1.0\text{-}1.1 \times 10^{-3}$) apparent diffuse coefficient (ADC) value and malignant PNSTs typically having a low ($<1.0\text{-}1.1 \times 10^{-3}$) ADC value [14].

Although histopathological diagnosis ultimately determines the final diagnosis, the presence of a large heterogeneous mass with markedly T2 hyperintense components intimately associated with the lumbosacral plexus in a patient with other imaging or clinical findings of NF-1, should raise the suspicion of a MPNST even if the tumor appears to have an intraosseous origin.

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