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Case Report

Leptomeningeal dissemination of a malignant melanotic nerve sheath tumor: A case report and review of the literature

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ABSTRACT

Background: Malignant melanotic nerve sheath tumors (MMNSTs) are rare tumors of presumed neural crest origin. Here, we present a 21-year-old female with a left L5/S1 MMNST along with a review of approximately 70 spinal cases reported in the literature, the majority of which were either local recurrences or metastases.

Case Description: A 21-year-old female presented with 3 months of severe left L5 distribution radicular leg pain and sensory loss. The MR revealed a dumbbell-shaped, heterogenously enhancing lesion centered on the left L5/S1 foramen; the intracanalicular component displaced the thecal sac to the right, while the extraforaminal portion of tumor extended anteriorly into the retroperitoneal space. Gross total resection was performed after a L5/S1 facetectomy. In the immediate postoperative period there were no complications, and the patient had full lower limb power. Four months later, the patient experienced generalized seizures, headache, and multiple cranial nerve palsies due to local and diffuse CNS dissemination. The MRI of the brain and whole spine revealed diffuse leptomeningeal enhancement along the full length of the spinal cord into the brainstem and cerebrum along with a focally recurrent epidural soft-tissue lesion located posterolaterally on the left at the L4/5 level (i.e., measuring 12 mm × 10 mm). An external ventricular drain and subsequent ventriculoperitoneal shunt were inserted, followed by craniospinal irradiation. She was discharged 3 months later with residual distal lower limb weakness.

Conclusion: This case illustrates the rapid disease progression of MMNST despite gross total resection. Further such lesions should be aggressively treated locally, and followed by adjuvant radiotherapy and systemic chemotherapy/immunotherapy.

Keywords: Adjuvant radiotherapy, Leptomeningeal dissemination, Malignant melanotic nerve sheath tumor, Melanotic schwannoma, Recurrence

INTRODUCTION

Malignant melanotic nerve sheath tumors (MMNSTs) of the spine are rare, and fewer than 70 cases are described in the literature.[5] According to the 5th Edition of the WHO classification of soft tissue and bone lesions, these most commonly affect the dorsal nerve roots but can also impact the entire neuroaxis and the gastrointestinal tract. Despite a relatively benign histological appearance, they exhibit high local recurrence rates and/or metastases. Here, we describe a 21-year-old female who originally presented with a

left foraminal L5/S1 dumbbell MMNST that recurred (i.e., locally at L5/S1) and metastasized (i.e., leptomeningeal spread) throughout the neuroaxis within just four postoperative months.

CASE REPORT

Original presentation and surgery

A 21-year-old female presented with 3 months of severe, left L5 radicular pain accompanied by L5 hypoesthesia to light touch and decreased pin-perception, without weakness. MR showed a dumbbell-shaped, heterogenously enhancing lesion centered in the left L5/S1 foramen, measuring 4.5 cm × 4 cm [Figure 1]. The tumor displaced the thecal sac anterolaterally to the right and also involved the posterolateral L5 vertebral body/left L5 pedicle/lamina along with superior extension (i.e., toward L4/5 into the retroperitoneal space above the sacral alar). Multiple tortuous vessels extended along the cauda equina [Figure 1].

Operation

Through a L4 and L5 hemilaminectomy with a left L5/S1 facetectomy, a gross total excision was accomplished. On opening of the dura, a black, hemorrhagic tumor was visualized. Postoperatively, the patient's radicular pain resolved, she retained full motor and regained normal sensory function in the left L5 distribution.

Local recurrence and leptomeningeal spread of tumor

Within 4 postoperative months and despite a negative PET-CT obtained 3 months after surgery, the lesion recurred. The patient presented with sudden onset lower back pain with recurrent radiculopathy. Within days, she also developed a severe bifrontal headache, vomiting, photophobia, visual loss (i.e., only able to differentiate between light and dark), and a generalized seizure. The patient also developed distal lower limb weakness, bilateral abducens nerve palsies, facial diplegia, bilateral trigeminal nerve palsies, and dysarthria. The fundus exam showed florid papilledema. The emergent holo-spinal and brain MRI showed diffuse leptomeningeal enhancement along the full length of the spinal cord extending to the brainstem and cerebrum. There was also a focal recurrent epidural soft-tissue lesion within the left posterolateral aspect at L4/5 measuring 12 mm × 10 mm [Figure 2].

A CT-guided lumbar puncture revealed only gelatinous material and core biopsy samples were taken. CSF studies showed high protein, low glucose, but no organisms. A frontal external ventricular drain was placed, and later converted to a ventriculoperitoneal shunt Craniospinal irradiation was administered after EVD insertion. Notably, the patient was discharged wheelchair-bound 3 months later.

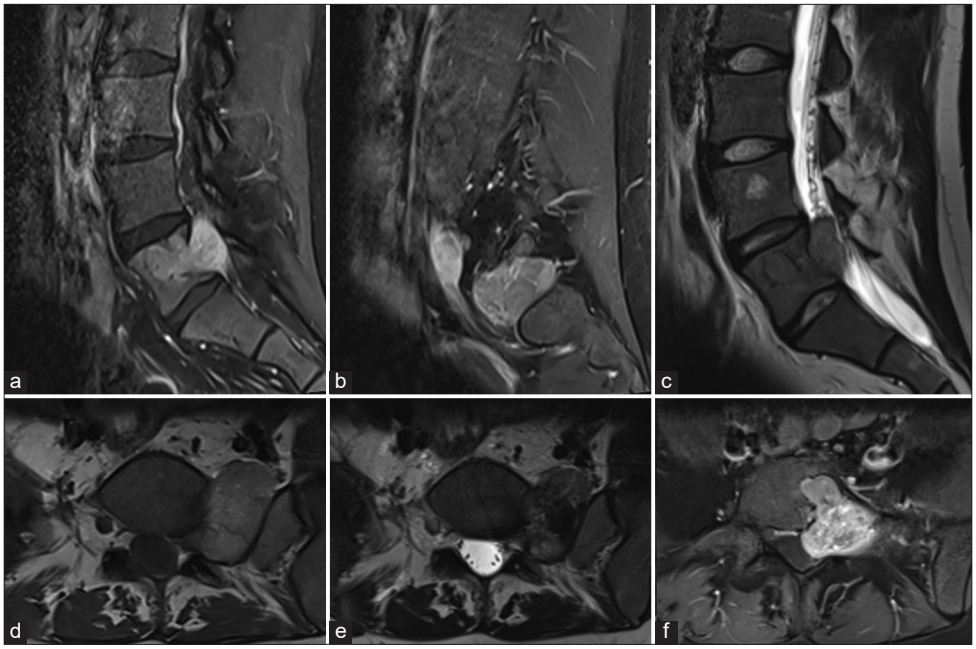

Figure 1: Preoperative MR images of the lesion. (a and b) Sagittal T1-weighted images with gadolinium enhancement of the lumbosacral spine demonstrating diffuse, heterogenous enhancement of the lesion with involvement the posterior aspect of the L5 vertebrae extending laterally to the retroperitoneal space. (c) Sagittal T2-weighted MR image showing extension of the lesion into the spinal canal and incidental hemangioma in the L4 vertebral body. (d) Axial T1-weighted MR image with lesion exiting the left L5/S1 foramen. (e) Axial T2-weighted MR image with lesion demonstrating low signal. (f) Axial T1-weighted image of the lesion with gadolinium enhancement showing heterogenous enhancement and rightward displacement of the thecal sac.

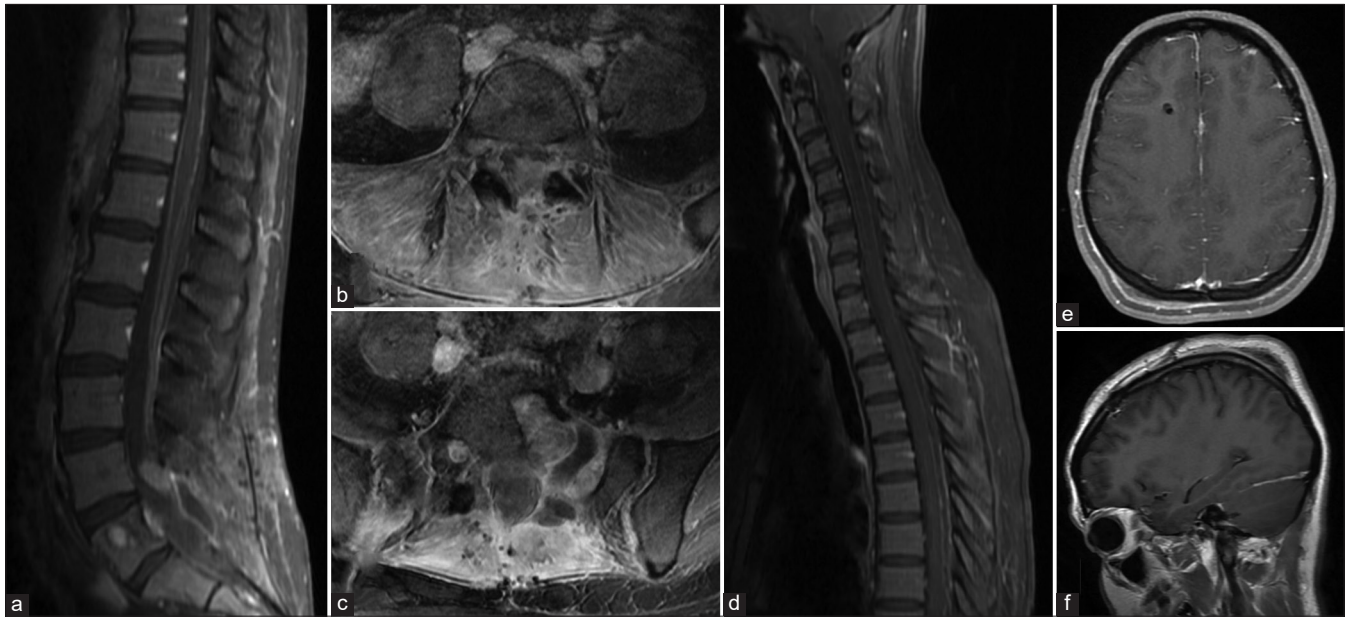

Figure 2: MR images of the brain and whole spine demonstrating local disease recurrence and diffuse supratentorial and spinal leptomeningeal enhancement. (a) Sagittal T1-weighted MR with gadolinium contrast image of the lumbosacral spine demonstrating enhancing focus at L4/L5. (b) Axial T1-weighted MR image showing enhancing focus. (c) Axial T1-weighted MR image showing tumor cavity. (d) Sagittal T1-weighted MR image with gadolinium contrast demonstrating diffuse leptomeningeal enhancement along the cervical and thoracic spinal cord. (e) Axial T1-weighted MR image with gadolinium contrast demonstrating dural enhancement along the anterior aspect of the falx cerebri. (f) Sagittal T1-weighted MR image with gadolinium contrast demonstrating possible dural enhancement along the right tentorium.

Histology

Tumor sections from both treatment periods showed a pigmented epithelioid to spindled cell tumor composed of cellular nodules, trabeculae and fascicles characterized by moderate nuclear pleomorphism, variably prominent nucleoli, and a moderate amount of eosinophilic cytoplasm [Figure 3]. The initial specimen had scattered nuclear grooves and intra-nuclear pseudo-inclusions, with only rare mitoses observed. No psammoma bodies were seen. The pigment was demonstrated to be melanin (Schmorl's positive, Perl's negative). The tumor cells were strongly positive for S100, HMB45, melanin-A and SOX10 and negative for EMA, AE1/3, CAM5.2, and GFAP. The proliferative rate was 6% (Ki67). These findings were consistent with a diagnosis of a MMNST. The secondary tumor showed MMNST, identical in morphology to that seen from the primary excision and included increased areas of tumor necrosis with a high mitotic count of up to 14 per high power field (×400). Further, the Ki67 had increased significantly and was estimated at 40% [Figure 4].

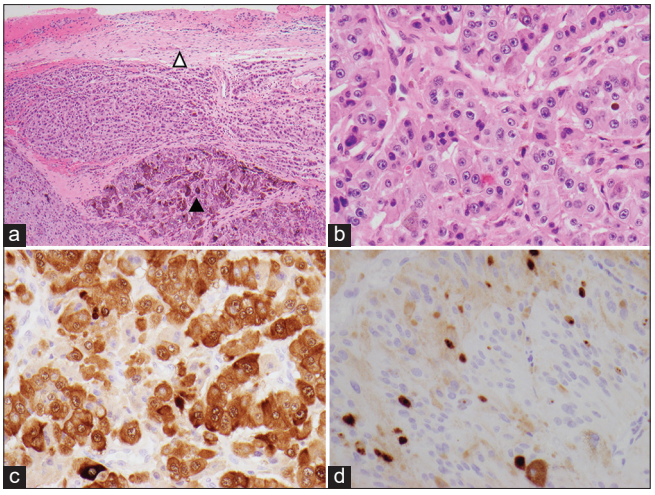

Figure 3: Photomicrograph of the specimen. (a) Pigmented epithelioid to spindled cell tumor surrounded by fibrous rim suggestive of dura (open arrowhead. Closed arrowhead demonstrating more heavily pigmented tumor) (H&E, ×40). (b) Moderate nuclear pleomorphism and variably prominent nuclei (H&E, ×400). (c) S100 immunostain with brown chromogen, nonpigmented area of tumor (×400). (d) Ki67% immunostain, brown chromogen (×400).

DISCUSSION AND LITERATURE REVIEW

MMNSTs are rare aggressive tumors of presumed neural crest origin which most commonly involve the dorsal spinal nerves but may affect the spinal cord, sympathetic chain, cranial nerves, the lumbar plexus, peripheral nerves, and the gastrointestinal tract. Approximately 200 cases have been reported in the literature since the first identified case published in 1931 by Millar, who described the

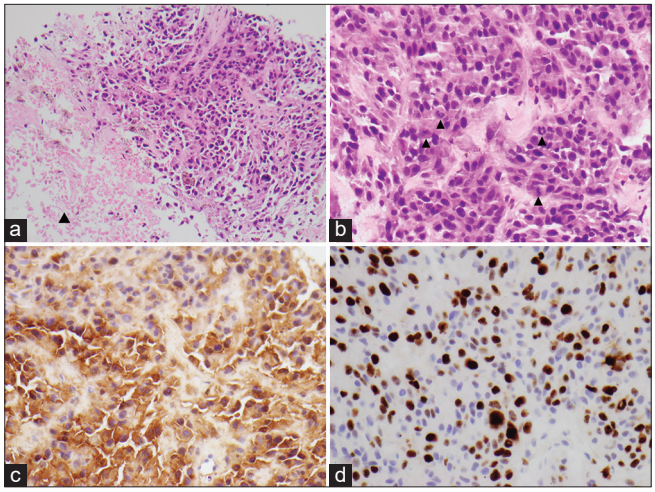

Figure 4: Photomicrographs of the core biopsy. (a) Arrowhead showing area of necrosis (H&E, ×200). (b) Arrowheads on mitotic figures (H&E, ×400). (c) S100 stain, brown chromogen (×400). (d) Ki67 approximately 40% (×400).

lesion as a "malignant melanotic tumor of the ganglion cells,"[4] with fewer than 70 of these lesions localized to the spine.[5] These lesions may be separated into psammomatous and non-psammomatous subtypes. Non-psammomatous lesions appear to arise sporadically, while psammomatous subtypes are considered to be part of the Carney complex, characterized by the presence of cutaneous lesions, endocrine tumors, and cardiac myxomas.[2]

These tumors were reclassified in 2020 by the WHO as "MMNSTs."[3] Reports have cited that 35% of patients experience local recurrences and 13–44% experience metastases.[6] A recent literature review found evidence of metastasis or local recurrence occurring in more than half of all cases.[5]

Diagnostic study of choice

MR imaging remains the imaging modality of choice for these tumors. The lesions typically appear hyperintense on T1-weighted images and hypointense on T2-weighted images.[5] CT of spinal nerve lesions often show enlarged focal intervertebral foramina with occasional adjacent bony erosion.[1]

Treatment recommendations

At present, there is no optimal treatment protocol for managing MMNSTs. Most studies advocate maximal safe resection. Radiotherapy is often offered only following local recurrence or distal metastases (i.e., for palliative symptom control). In this case, our patient did not originally receive adjuvant radiotherapy, as there was no evidence of residual tumor (i.e., following gross total resection). In this case and for patients with recurrent tumors or evidence of holo-spinal/brain leptomeningeal spread, craniospinal radiation with focal RT boosts to local lesions may be utilized.

CONCLUSION

MMNSTs are rare tumors that have a high propensity for local recurrence or metastasizing through the leptomeninges. Treatment recommendations include gross total initial resection followed by adjuvant radiotherapy and/or chemo-immunotherapy.

Declaration of patient consent

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

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Conflicts of interest

There are no conflicts of interest.

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