Intramedullary melanotic schwannoma: a rare presentation of a rare tumor. Illustrative case

Margaret M. McCann, MS,1,2 and John D. Hain, MD3

1Department of Translational Neuroscience, Barrow Neurological Institute, St. Joseph’s Hospital and Medical Center, Phoenix, Arizona; 2Department of Biomedical Sciences, Creighton University, Omaha, Nebraska; and 3Nebraska Spine Hospital, Omaha, Nebraska

BACKGROUND Intramedullary melanotic schwannomas (IMSs) are rare spinal cord tumors, and a hemorrhagic IMS is an even rarer presentation of this uncommon lesion. The authors describe the second known case of a hemorrhagic IMS and briefly review the characteristics of IMSs.

OBSERVATIONS The patient’s initial presentation and imaging indicated an intramedullary thoracic spinal cord tumor impairing lower extremity function. Intraoperatively, the lesion appeared pigmented and hemorrhagic. Pathologic analysis determined the tumor to be an IMS.

LESSONS Melanotic schwannomas vary in presentation and can resemble malignant melanoma but are differentiated by pathologic markers. Lesions typically present as extramedullary masses in the thoracic cord. Intramedullary presentation is rare but should be considered for pigmented tumors.

https://thejns.org/doi/abs/10.3171/CASE22391

KEYWORDS intramedullary spinal cord tumor; malignant melanotic nerve sheath tumor; melanocytic schwannoma; neurilemmoma; spinal cord neoplasm

Melanotic schwannomas (MSs) are uncommon, poorly understood nerve sheath tumors. Intramedullary MSs (IMSs) are even rarer lesions, with only 12 cases of IMSs reported in the literature as of 2022.1–12 Hemorrhagic IMSs are rarer still, and here we present a case report describing only the second recorded instance of hemorrhagic IMS (the first was in 2014).8

In 2016, MSs were reclassified as their own malignant type of schwanna due to their greater malignancy than ordinary schwannomas.13 However, why a peripheral nerve tumor may present in the intramedullary space is still unknown. The driver of hemorrhagic conversion in these tumors is also unknown, and we do not yet know how that conversion affects progression and prognosis. Case reports such as this help characterize a poorly understood tumor and add to the growing body of literature, which will aid in the understanding and treatment of these rare spinal cord tumors.

Illustrative Case

Presentation and Imaging

A man in his early 40s presented with a 2-month history of progressive bilateral leg weakness and pain that he described as sharp, shooting, and pressure-like. The pain, which was aggravated by activity, began in his back and radiated bilaterally to his lower extremities. He had associated paresthesia. At the time of presentation, he used a wheelchair and had difficulties with bladder and bowel functions. A review of systems was unremarkable except for abdominal pain and sleep disturbance. The patient's medical and surgical histories were remarkable only for a previous hernia repair. The patient reported marijuana use but denied any history of tobacco, alcohol, or other drug use.

Upon examination, the patient was hypertensive with otherwise normal vital signs and a body mass index of 25 kg/m2. A thorough neurological examination found decreased strength of the lower extremities in both flexion and extension of the hips, knees, and toes. This weakness was more pronounced on the left than the right, with corresponding bilateral hyperreflexia of the patellar and Achilles reflexes.

Magnetic resonance imaging (MRI) of the thoracic and lumbar spine showed an inherently T1-enhancing intramedullary mass extending from T8 to T11 (Fig. 1). On MRI, the lesion measured 3.1 cm cephalocaudally with a maximal visible diameter of 11 mm. The mass was noted to be cystic and septate with hemorrhagic components.
Additionally, a septate syrinx, measuring 12 mm at its widest, was noted to extend the length of the thoracic cord. Urgent surgery was planned.

**Surgery and Diagnosis**

A T9–12 laminectomy was performed, followed by durotomy of the corresponding levels. Upon exposure, the cord appeared darkened from subacute hematoma, with visible pockets of fresh blood. A midline myelotomy was started above and below the lesion and was worked back toward the lesion. At this point, the hematoma section of the tumor self-evacuated, and a steady flow of mild to moderate bleeding was observed around the tumor periphery. The lesion itself was jet black and appeared more destructive than an ependymoma, immediately raising suspicion of malignant melanoma. A friable capsule was encountered, which allowed clean dissection. A large vascular supply was located inferiorly along the length of the tumor, extending from the T10–11 disc space through the T11–12 disc space. Despite meticulous dissection, the lesion bled steadily throughout the procedure. Eventually, the bulk of the tumor and the loculated hematoma were removed. By the end of the resection, motor evoked potentials to the sphincter, bladder, and left lower extremity (LLE) were undetectable, although some response in the right lower extremity remained. The resection could not be stopped sooner because portions of the tumor and hematoma had erupted from the cord, and the dura could not be closed over the autoevacuation. Furthermore, given the amount of bleeding seen from the periphery of the tumor, the cord could not be closed until bleeding was controlled. Once the bleeding was controlled, surgical resection and further exploration were halted because of the loss of motor signals; however, visual inspection indicated that no residual tumor remained.

**Pathology**

Preliminary intraoperative pathology reports were strongly suspicious for malignant melanoma. Postoperatively, permanent sections demonstrated heavily pigmented epithelioid to spindled cells arranged in nests and cords with some lymphocytic infiltrate but no psammoma bodies (Fig. 2). Cells had enlarged ovoid nuclei with irregular nuclear contours, variably cherry red nucleoli, and gray cytoplasm. Only 1 to 2 mitoses per 10 high-power fields (HPFs) were observed. Table 1 lists molecular pathology targets and their significance. In this case, the specimen was positive for SOX10 (Sry-related HMg-box gene 10) and melanoma antigen recognized by T cells (Melan-A), consistent with a melanocytic lesion. The Ki-67 antigen, which marks the Ki-67 protein proliferation index, was not significantly increased, and proto-oncogene B-Raf was negative, decreasing the likelihood of melanoma. Histological features indicated a malignant melanotic nerve sheath tumor, and this diagnosis was confirmed by the Cleveland Clinic.

**Postoperative Evaluation**

Postoperatively, the patient required approximately 3 weeks of inpatient care and rehabilitation. During this time, his LLE proximal muscle strength increased. At the time of discharge, the patient had light touch sensation to his LLE and was able to internally rotate his left hip well. Other motor functions on the proximal LLE started to show improvement. The patient’s right lower extremity was weak throughout and lacked sensation and proprioception, but he was able to raise it against gravity. While hospitalized, the patient was medically stable and maintained bladder and bowel function. An MRI performed 1 month after the operation showed gross total resection of the tumor, and additional imaging showed no discernible metastasis to other parts of the central nervous system, chest, abdomen, or pelvis.
Schwann cell extension from dorsal root insertion sites. Although dered neural crest migration during embryogenesis to neoplastic these tumors develop is unknown, but theories range from disor-

ginal nerve sheath tumors” when referring to MSs, to better align with terminology used in soft-tissue patholo-
gle of origin.7 MSs were separated from other schwannomas and reclassi-

ed as their own type of malignant tumor in 2016.13 Of note, the 2021 World Health Organization Classification of Tumors of the Central Nervous System suggests the use of the phrase malignant to be a simple schwannoma or ependymoma, and the gross appearance of the lesion suggested that it was too malignant containing melanin15

discriminated neural crest migration during embryogenesis to neoplastic Schwann cell extension from dorsal root insertion sites.7 Although MSs were originally thought to derive from rostral components of the neural tube, a 2008 report of an isolated tumor of the conus medullaris called that theory into question, and their exact etiology remains undetermined.5,21 Our patient has been used for extramedullary spinal tumors with some suc-

cient residual back pain. On strength testing, he showed antigrav-

derived from nerve sheath cells, MSs are differentiated from schwannomas in part by their melanin expression and lack of glial fibrillary acidic protein staining. In our case, the radiological and pathologic evidence was consistent with previous reports. Preopera-

tively, the radiological appearance of the lesion showed a cystic, partially enhancing, hemorrhagic mass that could be indicative of aggressive course, and because of the extension of the tumor and extension of the tumor and intraoperative expansion caused by the hemorrhage, adjuvant radio-

TABLE 1. Pathology markers and their significance

<table>
<thead>
<tr>
<th>Name</th>
<th>Abbreviation</th>
<th>Function</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sry-related HMg-box gene 10</td>
<td>SOX10</td>
<td>A nuclear transcription factor important in melanocytic cell differentiation14</td>
</tr>
<tr>
<td>Melanoma antigen recognized by T cells</td>
<td>Melan A</td>
<td>A melanosomal differentiation antigen expressed by healthy &amp; malignant tissue containing melanin15</td>
</tr>
<tr>
<td>Ki-67 antigen marks Ki-67 protein</td>
<td>Ki-67</td>
<td>A protein present during active cell phases (G1, S, G2). Associated w/ cell proliferation in malignant tumors. Used as marker of tumor aggressiveness.16</td>
</tr>
<tr>
<td>B-Raf proto-oncogene</td>
<td>BRAF</td>
<td>Mutation present in 90% of malignant melanomas, commonly negative in melanotic schwannomas. Activates a proliferation pathway called “mitogen-activated protein kinase pathway.”17,18</td>
</tr>
<tr>
<td>Glial fibrillary acidic protein</td>
<td>GFAP</td>
<td>Intermediate filament protein used to identify astroglial cells19</td>
</tr>
</tbody>
</table>

At 3-month follow-up, the patient continued to use a wheelchair but had continued slow improvement in motor function, and he reported no significant residual back pain. On strength testing, he showed antigravity strength in the proximal LLE, severe continued paresis of the distal LLE, and 4/5 on the right lower extremity, although he still complained of decreased sensation bilaterally in his legs. He was scheduled for continued occupational and physical therapy, plus radiation treatment as recommended by the oncologist.

Discussion

Observations

IMSs are rare, with only 12 cases previously reported. How these tumors develop is unknown, but theories range from disor-

s to better align with terminology used in soft-tissue patholo-
gic imaging, reflecting the underlying variability of the tumors them-
selves. The pathologic features of this lesion were consistent with features of previously reported MSs. In this case, the MS demonstrated the mixed features of a melanocytic lesion that was also positive for nerve sheath tumor markers. In addition to the patholog-

cell shape, greater nuclear pleiomorphism, and lower mitotic morphological appearance because MSs have a more spindled rate than melanomas. Careful dermatologic examination to rule out melanoma is crucial to rule out metastatic disease. Although derived from nerve sheath cells, MSs are differentiated from schwannomas in part by their melanin expression and lack of glial fibrillary acidic protein staining. In our case, the radiological and pathologic evidence was consistent with previous reports. Preopera-

tively, the radiological appearance of the lesion showed a cystic, partially enhancing, hemorrhagic mass that could be indicative of several tumor types, including ependymoma, melanoma, or MS. As noted by Alamer and Tampieri,12 MSs are variable in their radio-

aggressiveness.16

Prognosis is difficult to determine for patients with MSs because of the relative rarity of these tumors and the variability of their pre-
sertation. A higher mitotic rate (=2 mitoses per 10 HPFs) has been noted to correlate with metastasis, but the converse is not necessarily true, and benign-looking lesions still have malignant protentional.19 To date, no markers have been identified that allow definitive predictions.12,19,21

No consensus exists regarding the use of adjuvant radiotherapy for MS, although some studies argue that adjuvant radiotherapy is beneficial for intracranial MSs.22–24 Scant data exist to support adjuvant radiotherapy in the intramedullary spinal variety, although it has been used for extramedullary spinal tumors with some success.5,21 Our patient’s tumor had a relatively low mitotic rate (2 mit-

oses per HPF), but, as noted above, low mitotic rate does not predict less malignancy. The rapid onset of symptoms indicated an aggressive course, and because of the extension of the tumor and intraoperative expansion caused by the hemorrhage, adjuvant radio-

therapy was included in the treatment plan.
Lessons

MSs are rare tumors of the spinal cord. We present only the second reported case of a hemorrhagic IMS. It is important to recognize these tumors because of their rapid progression and malignant potential. More data are needed to determine the pathophysiology and prognosis of IMS and to help define treatment guidelines.

Acknowledgments

We thank the patient for his help and cooperation in sharing his case. Additionally, we thank Dr. Steven Billings of the Cleveland Clinic for his help with the pathology analysis and for providing a definitive diagnosis; Robin Carlson, MSN, of Nebraska Spine Hospital for her invaluable help compiling resources; and Drs. Wayne Penka and Joyce Kover for providing the pathology image. We thank the staff of Neuroscience Publications at Barrow Neurological Institute for assistance with manuscript preparation.

References


Disclosures

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author Contributions

Conception and design: Hain. Acquisition of data: McCann. Analysis and interpretation of data: McCann. Drafting the article: McCann. Critically revising the article: both authors. Reviewed submitted version of manuscript: both authors. Administrative/technical/material support: Hain. Study supervision: Hain.

Correspondence

Margaret M. McCann: c/o Neuroscience Publications, Barrow Neurological Institute, St. Joseph’s Hospital and Medical Center, Phoenix, AZ. neuropub@barrowneuro.org.

4 | J Neurosurg Case Lessons | Vol 5 | Issue 7 | February 13, 2023