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(CASE REPORT)

Recurrent Metastatic Malignant Peripheral Nerve Sheath Tumor with Complicated Clinical Course: A Case Report and a Brief Review of the Literature

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Abstract

Malignant peripheral nerve sheath tumor (MPNST) is a rare, high-grade soft tissue sarcoma that poses substantial diagnostic challenges. This tumor poses significant diagnostic challenges due to variable histologic patterns, limited neural marker expression, and overlap with the spectrum of other high-grade sarcomas. Its heterogeneous histologic patterns, limited expression of neural markers, and significant morphologic overlap with other high-grade sarcomas often complicate accurate identification. Diagnosis is challenging and often requires contributions from clinical findings, radiology, immunohistochemistry, and molecular diagnostics, necessitating a multidisciplinary team for diagnosis and management. MPNST is associated with a high rate of recurrence and metastasis; therefore, early detection and lifelong follow-up after treatment are essential. Here, we report a 39-year-old woman who was diagnosed with extraskeletal myxoid Chondrosarcoma (EMC) after excision of a left thigh mass. Two years afterward, she had locally extensive recurrence and FDG-avid mediastinal metastases. Advancing quality-assessment, comprising extensive histopathology and focused molecular investigations, reclassified the diagnosis as high-grade MPNST, based on CDKN2A deletion and the absence of NR4A32 rearrangement. This clarification changed the management, referring her to neoadjuvant chemotherapy, debulking surgery, and a subsequent plan of adjuvant therapy. This revised diagnosis prompted a change in therapeutic strategy, with initiation of neoadjuvant chemotherapy followed by surgical debulking and planned adjuvant therapy. Although she had an initial response, the patient developed widespread metastatic disease and died 8 months later. This case highlights the difficulties in diagnosing MPNST, the importance of interdisciplinary teamwork, and the need for close monitoring in patients with high-grade MPNST.

Keywords: Peripheral nerve sheath tumor; Malignant; Epithelioid; Extraskeletal myxoid chondrosarcoma; Reclassification; Molecular

1. Introduction

MPNST is a rare and aggressive soft tissue sarcoma of neural origin, which represents < 5% of soft tissue sarcomas. [1] It has a dismal prognosis due to the high probability of local recurrence and distant metastasis. MPNST is predominantly seen in patients with NF1 and rarely occurs sporadically, as in the patient in our report. Management of MPNST is challenging, and aggressive surgical resection with adjuvant radiotherapy and systemic chemotherapy in a multidisciplinary manner can be offered, yet the overall survival remains very poor. [2]

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Diagnosing MPNST is one of the challenging diagnoses for surgical pathologists. [3] The tumor displays significant histological heterogeneity, where it can morphologically and immunophenotypically (IHC) overlap with other high-grade spindle cell sarcomas such as EMC, synovial sarcoma, and leiomyosarcoma. [4] High-grade or dedifferentiated MPNST loses expression of S-100 protein further adding to diagnostic confusion by removing the historically valuable marker for neural lineage. The diagnostic uncertainty of this lesion underscores the essential role of sophisticated molecular and IHC studies to establish the correct diagnosis and provide optimal management. [4]

We present a complicated case of recurrent metastatic high-grade MPNST in a 39-year-old female, initially misdiagnosed as EMC following the excision of a primary thigh mass. The development of extensive local recurrence and FDG-avid mediastinal metastases prompted a comprehensive quality-assurance review of the original pathologic diagnosis. Expanded immunohistochemical analysis and advanced molecular testing led to reclassification of the tumor as high-grade MPNST, based on identification of CDKN2A deletion and the absence of NR4A3 rearrangement. This correction considerably changed the strategy for the patient's treatment towards neoadjuvant chemotherapy and debulking surgery. Despite an initially favorable response to aggressive multimodality treatment, the patient experienced rapid disease progression and died within eight months of the revised diagnosis.

2. Case Presentation

2.1. Primary presentation and history of prior treatment

A 39-year-old female patient reported to the oncology clinic with multifocal recurrent disease and mediastinal metastases two years following resection of a tumor in the thigh that was initially diagnosed as EMC. Two years prior to the current presentation, the patient had a 9 cm left anterior thigh mass in the femoral triangle resected. The tumor was closely connected with the femoral nerve. The initial diagnosis was EMC. The surgical margins were negative, but one of them was less than 1 mm. Adjuvant radiotherapy was advised, but the patient refused additional treatment. The primary surgery and skeletal survey showed no evidence of metastasis.

2.2. Current Presentation

Currently, the patient presented with progressive swelling and pain of the left thigh and signs associated with mediastinal compression. Radiography showed a recurrent 12 x 9 cm mass in the thigh at the former surgical site, and three mediastinal masses, the largest measuring 3 cm. No other disease localization was found. MRI and CT of the thigh revealed a complex mass with characteristics suggestive of aggressive behavior, including areas of necrosis and hemorrhage. Both the thigh and mediastinal lesions were hypermetabolic, with intense hypermetabolic activity evident on FDG-PET.

2.3. First Tumor Board Review

A multidisciplinary tumor board convened to strategize the optimal approach, debating between immediate resection of the thigh mass or performing a tissue biopsy to define the diagnosis. The diagnosis of the current mass necessitated consideration of the possibility of recurrence of the same tumor type, which led the board to suggest a biopsy of the recurrence and a complete re-evaluation of the initial pathology. This was aimed at creating a diagnosis and setting the order of treatment, including potential neoadjuvant therapy before surgery.

2.4. Pathologic Review of the Primary Tumor and Molecular Studies

Review of the previous tumor slides (two years ago) showed a high-grade sarcoma with heterogeneous morphology, including angiosarcoma-like, myxoid, chondroid-like, and epithelioid regions. Synaptophysin, neurofilament protein, Neu-N, S-100 (weak, focal), and low molecular weight cytokeratin CAM 5.2 had been positively expressed by IHC. There was also focal positivity of AE1/AE3. Ki-67 labeling approached 35%. INI-1 expression was retained. The tumor did not express CD31, CD34, Factor VIII, myogenic markers, or melanocytic markers, including HMB45.

2.5. Pathology of the Recurrent Tumor

Microscopic examination of the biopsy of the recurrent thigh mass revealed features of epithelioid MPNST with a nodular pattern separated by fibrous septae. The tumor cells had small, round to epithelioid morphology, with significantly increased nuclear atypia and a mitotic rate exceeding 20 per 10 high-power fields. A focal myxoid background was noted. This indicated that the lesion has progressed to a more advanced phenotype. (Figure 1 A, B, C) There was a positive result on IHC of vimentin, SOX10, CD68, synaptophysin, CD56, CD57, and CD117, and a negative result of CK20 and S-100. (Figure 2 A, B, C, D) Negative reaction was also observed with the following markers: all cytokeratins, CD99, Desmin, Myogenin, CD34, and melanocytic markers. The pathologists agreed that the initial and

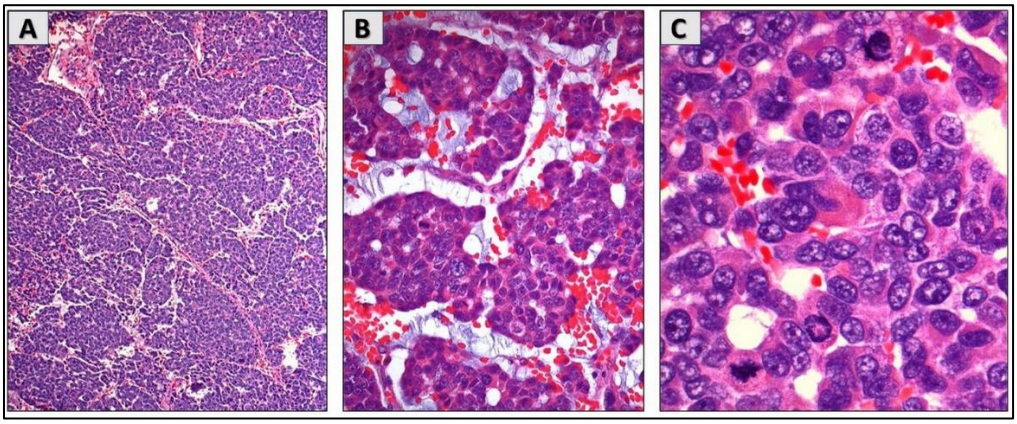
recurrent tumors are the same. S-100 was lost due to dedifferentiation, but because of the changing morphology and immunoprofile, they proposed molecular studies to clarify these changes.

Because it was still necessary to differentiate between EMC and MPNST as part of the diagnostic process, FISH studies were performed on both tumors, the initial and the recurrent tumors. Negative NR4A3 rearrangement in FISH ruled out the diagnosis of EMC. [5] FISH demonstrated that it had lost CDKN2A, which is associated with MPNST. [6] H3K27me3 is an immunohistochemical stain that is now routine in MPNST evaluation, but was not performed at the time of the evaluation. [5] [6] The patient's tumor was finally reclassified as a high-grade MPNST based on molecular findings, histopathology, and clinical behavior.

2.6. Management and outcome

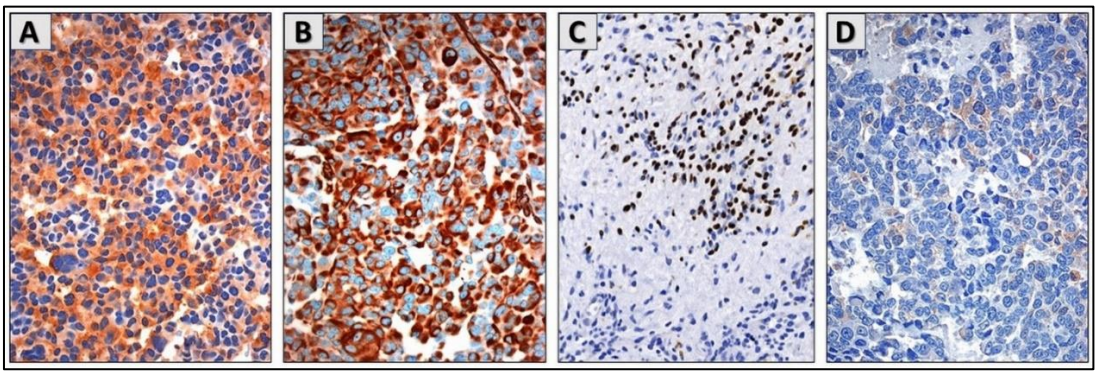
The patient began neoadjuvant chemotherapy with ifosfamide and doxorubicin. She responded favorably, most of her symptoms were alleviated, and there was a partial decrease in the tumor load. Surgical debulking of the thigh mass and all available metastatic mediastinal masses was performed without significant complications. Postoperative adjuvant chemotherapy and radiation therapy were initiated. The same chemotherapy as the neoadjuvant therapy was used. However, the patient suffered from severe treatment-related toxicity, resulting in multiple interruptions and discontinuation of adjuvant therapy. Even with attempts at systemic control, she continued to develop disease progression.

The patient developed progressive metastases and functional decline over the next 8 months. She deteriorated with disease progression despite supportive care and a series of treatment modifications and succumbed to disseminated MPNST.



1A Low power view showing features of epithelioid MPNST with nodular pattern separated by fibrous septae (H&E stain X20)' 1B Intermediate power view showing focal myxoid background (H&E stain X40); 1C High power view showing pleomorphic, highly atypical tumor cells with frequent mitosis (H&E stain X60)

Figure 1 Recurrent tumor (MPNSH) microscopic features



2A Tumor cells positive for vimentin; 2B Tumor cells positive for Synaptophysin; 2C Tumor cells positive for SOX10; 2D Tumor cells negative for S-100 (Dedifferentiation)

Figure 2 Recurrent tumor (MPNST) immunohistochemistry studies

3. Discussion

3.1. Background (History, epidemiology, and WHO classification)

In 1982, R. A. Erlandson and J.M. Woodruff were the first to describe MPNST, when they used electron microscopy to visualize the ultrastructural appearances of peripheral nerve sheath tumors. [7] Their study established the characteristic criteria of MPNST histology and helped to differentiate it from other nerve sheath tumors, thus defining a group that was previously largely unrecognized.

The results of the patient's treatment were generally unsatisfactory because of the tumor's aggressive nature, high recurrence rate, and extensive metastasis. Recently, there has been a focus on molecular and genetic drivers of correct MPNST diagnosis, with emphasis on biomarkers such as NF1, TP53, and changes in the RAS/RAF/MEK/ERK signaling cascade, which play vital roles in tumorigenesis. [8]

Epidemiologically, MPNST is a rare but highly aggressive soft tissue sarcoma that accounts for approximately 5-10% of all sarcomas. For localized MPNST, the 5-year survival is poor, approximately 30%-60%, but falls dramatically if a metastasis or recurrence occurs, with less than 7-10% in high-grade disease. [9] It is associated with a significantly increased risk of development in patients with a history of NF-1, with an estimated risk of 8-13%. [10] In these patients, bi-allelic mutation of NF-1 is necessary for the development of MPNST, but it is not sufficient. For full malignant transformation, mutations in genes such as CDKN2A, EGFR, SUZ12, and TP53 are required. [11]

Major metastatic sites of MPNST are the lungs, bone, and liver, which makes these conditions one of the most lethal sarcomas. [12] Advancement in diagnostic resources has aided in distinguishing MPNST from other high-grade spindle and epithelioid cell sarcomas, and it has been shown that the unique genetic alterations drive aggressive and metastatic behavior. [13]

In 2013, the World Health Organization (WHO) first classified MPNSTs as soft tissue sarcomas. [1] The specific subtypes were described based on their cellular features, including epithelioid and malignant triton tumors. These tumors have been graded II, III, and IV based on the degree of differentiation. [14] The 2013 and the current 2020 WHO editions describe MPNST as a malignant spindle cell neoplasm arising from peripheral nerves and neurofibromas. However, the 2020 edition specifically classified MPNST on a molecular basis to distinguish it from conditions such as synovial sarcoma, other spindle cell sarcomas, and spindle cell melanoma. NF-1, CDKN2A, and PRC2 complex mutations were required to make the specific diagnosis of MPNST. [14] [15]

3.2. Pathogenesis, pathophysiology

MPNST is an extremely aggressive soft tissue sarcoma that differentiates into cells of the nerve sheath. It is generally believed that the cell of origin is the Schwann cell or a pluripotent precursor cell of the peripheral nerve sheath. [1] The etiology of MPNST is marked by the successive changes in genes and molecular structures that propel the malignant change. The most frequent genetic event is the loss of the Neurofibromin 1 (NF1) tumor suppressor gene, which is present in more than half of cases and often results in overactivation of the Ras signaling pathway. [5] [16] Nonetheless, in sporadic MPNST, which is the case in this case, loss of other critical tumor suppressors is the rule. [5]

One of the key changes in this patient was the loss of the CDKN2A gene, which is found in about 81 percent of MPNSTs. [6] CDKN2A encodes two tumor suppressors, p16INK4a and p14ARF, which regulate the cell cycle pathways of retinoblastoma (Rb) and p53, respectively. CDKN2A loss leads to uncontrolled cell division and is a key driver of malignant progression and high-grade transformation. [13] These molecular aberrations are a direct cause of the aggressive growth and spread of the tumor as it invaded the mediastinum, after starting at the primary location in the thigh and having a close relationship with the femoral nerve. Histopathologic correlates of this high-grade dedifferentiated phenotype are the high mitotic rate (>20 per 10 high-power fields) and loss of S-100 expression in the recurrent tumor. [1] Medically, the location and growth of the tumor resulted in the local symptoms of progressive swelling and pain because of mass effect on the surrounding tissues and nerves, as well as the mediastinal metastases, resulting in the symptoms of compression of vital structures.

More recent studies reported the loss of trimethylation at lysine 27 on histone H3 (H3K27me3) as strongly indicative of MPNST evolution. [17] Tumors that are characterized by H3K27me3 loss typically harbour large-scale alterations, including genome doubling and chromosomal loss, and can be detected using chromosomal structure analysis and cell-free DNA profiling, which is especially valuable in establishing a strong diagnosis. [18] From extensive studies on transcriptomic subtyping, the absence or presence of H3K27me3 defines various compliant MPNSTs subgroup

structures with distinct outcomes. [18] This suggests that the integration of IHC (e.g., S-100, SOX10, and H3K27me3), morphology, and genome structures of CDKN2A and PRC2 is representative of a strong basis for accurate diagnosis.

3.3. Comparative analysis of our case with the existing literature.

The clinical course of this 39-year-old woman with a recurrent, high-grade epithelioid MPNST demonstrates several interesting similarities and differences to those previously reported for this uncommon malignancy. [19] The patient's age of 39 years at presentation is consistent with the typical sporadic MPNST, which most commonly presents between the ages of 20 and 50 years. [1] Although 50% of MPNST cases are associated with NF-1, a Sporadic pattern is also observed, which is itself very aggressive. [16] [22] The site of the tumor in the left anterior thigh, adjacent to the femoral nerve, is one of the common locations for MPNST, and proximity to a major nerve is known to be an adverse prognostic indicator. [1] [20]

In our case, the recurrent tumor maintained the general epithelioid characteristics of its original type, consistent with the reported literature. [21] Epithelioid MPNST is an uncommon, highly aggressive variant and more often unrelated to NF1. [21] The aggressive biology was also supported by the molecular results, including proven inactivation of the CDKN2A tumor suppressor. This change was associated with a high-grade, aggressive course of disease and poor prognosis; the finding has been observed in the vast majority of MPNST. [6] [13] The occurrence of local recurrence and mediastinal metastases (which occurs in only 10% of the patients upon initial presentation) suggested a very aggressive biology with an expected worse prognosis. [22] [23]

The primary misdiagnosis with EMC can be the most important alternative diagnosis. MPNST is a great imitator, and the heterogeneous morphological appearance of the primary tumor, which contained myxoid and epithelioid areas, likely contributed to difficulties in correct diagnosis. The confirmatory diagnosis necessitated molecular studies (FISH for NR4A3 rearrangement and CDKN2A loss), which are increasingly recognized as a critical adjunct for correctly classifying diagnostically difficult sarcomas. [6]

The treatment strategy, neoadjuvant chemotherapy followed by surgical debulking, was consistent with the high-grade, locally advanced sarcoma standard of care to downstage the tumor, as well as evaluate tumor chemosensitivity, [7] the partial response to ifosfamide and doxorubicin warranted further surgery. However, patient failure to complete the intended adjuvant therapy in the setting of significant toxicity, along with aggressive biology evidenced by CDKN2A loss and early metastasis, ultimately resulted in a quick and unfortunate course. [6] In our case, the patient expired from the disease 8 months after the recurrence and metastasis, a course which is strikingly in contrast to the reported 5-year survival rate of 42%–50% for sporadic non-metastatic MPNST. This emphasizes that MPNST remains a high-risk, difficult-to-treat oncologic entity once it has metastasized and demonstrates these high-risk molecular features. [1]

4. What have we learned from this case?

This case of recurrent, metastatic MPNST offers key diagnostic and therapeutic lessons that support current understanding around sarcoma oncology. The most important diagnostic teaching point is that diagnosis of high-grade sarcomas is an interdisciplinary, iterative process. The initial misdiagnosis of EMC, despite the tumor's proximity to a large nerve, illustrates the diagnostic challenge of MPNST, especially its epithelioid counterpart. This case further demonstrates the importance of: 1) Pathology re-evaluation of all primary specimens for recurrence or metastasis, and 2) Standard molecular testing to rule in or out specific sarcoma subtypes. The application of FISH in excluding EMC and confirming CDKN2A loss proved vital for the proper diagnosis, management, and prognosis. Additionally, the case emphasizes the need to incorporate newer, highly specific IHC markers in MPNST evaluation, thereby enabling earlier diagnostic clarity. [20] [24]

For clinicians, red flags to consider include: any rapidly growing mass associated with a significant nerve, and high-grade sarcoma with an indeterminate immunoprofile. The management and surveillance implications here are for those patients with high-grade MPNST or those with molecular markers of aggressiveness, such as loss of CDKN2A expression, who need aggressive long-term surveillance and have a low threshold for systemic therapy despite close margins. This case serves as a reminder that despite aggressive multimodal therapy, the outcome for metastatic MPNST remains exceedingly grim, and there is an ongoing need for further investigation into new targeted agents.

Abbreviations

Malignant peripheral nerve sheath tumor (MPNST);
Extraskeletal myxoid chondrosarcoma (EMC);

Immunophenotypically (IHC)

Conclusion

This case is presented to highlight the diagnostic challenges and clinical aggressiveness of MPNST, which can exhibit morphologic features that mimic those of other sarcoma subtypes. The initial misclassification of our patient's tumor for extraskeletal myxoid chondrosarcoma illustrates that morphology and even IHC may yield an incomplete diagnosis. It emphasizes the need for a multidisciplinary approach, including molecular investigations, to achieve an accurate diagnosis. The care team reclassified the diagnosis, and the patient received an appropriate course of treatment based on a reassessment of the primary tumor, with reference to detailed IHC studies and updated genetic information.

We present this case to raise the medical community's attention to pitfalls in diagnosing high-grade sarcomas, including the importance of shared decision-making, complete pathologic review, and long-term clinical follow-up. Early detection of MPNST and proper classification might assist in better treatment planning and, ultimately, an efficient treatment outcome for the patient.

Compliance with ethical standards

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Disclosure of conflict of interest

All authors make the following declarations:

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- Financial relationships: All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might be interested in the submitted work.

Statement of ethical approval

Ethical review and approval were not required for this study involving human participants. The paper has been sufficiently anonymized to maintain the patient's confidentiality.

Data access statement

All relevant data are included in the paper.

Author contributions

All authors contributed equally to producing this manuscript.

Statement of informed consent

The patient expired, and all attempts to reach the family members were unsuccessful. Therefore, the paper has been sufficiently anonymized to maintain patient confidentiality.

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