Case Report

CT of malignant peripheral nerve sheath tumor✩✩

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ARTICLE INFO

Article history:
Received 27 October 2022
Accepted 30 October 2022

Keywords:
Malignant peripheral nerve sheath tumor
Neurofibromatosis type 1
Neurofibromin 1 gene
Computed tomography

ABSTRACT

Malignant peripheral nerve sheath tumors are soft tissue sarcomas that typically arise from a neurofibroma. Patients with neurofibromatosis type 1 represent approximately half of the population diagnosed with these tumors. This autosomal-dominant genetic disorder is distinguished by loss-of-function mutations in the neurofibromin 1 gene, which ultimately promotes atypical cellular proliferation. These biologically aggressive tumors are associated with a poor prognosis as they are resistant to available therapies and have high rates of recurrence, progression, and mortality. In this article, we report the case of a 45-year-old male with a history of neurofibromatosis type 1 who was diagnosed with a malignant peripheral nerve sheath tumor. We focus on optimizing diagnosis and treatment through the application of radiological imaging modalities, including cinematic rendering.

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Introduction

Malignant peripheral nerve sheath tumors (MPNST), which arise as a result of associations with a peripheral nerve, account for approximately 5%-10% of all soft tissue sarcomas [1–3]. While an estimated 50% of all MPNSTs originate sporadically, the other half develop in the context of neurofibromatosis type 1 (NF-1), an autosomal-dominant genetic disorder characterized by mutations in the neurofibromin 1 gene (NF1) [4]. Affecting 1 in 3000 live births, NF-1 is characterized by distinct neurofibromas and various areas of cutaneous hyperpigmentation, also known as café-au-lait macules [5]. Patients have a 60% risk of developing cancer throughout their lifetime, with MPNSTs being the leading cause of death [6,7]. More specifically, the prevalence of MPNST in patients with NF-1 is 1:3500, a staggering difference from the incidence of 1:10,000 seen in the general population [8]. Thus, new-onset pain in the neurofibromas of NF-1 patients should be evaluated for MPNST [1]. Ultimately, poor prognosis, resistance to current therapies, and high rates of progression, recurrence, and mortality render management and treatment of MPNSTs extremely challenging.

✩ Funding: There was no funding associated with this work.
✩✩ Competing Interests: None of the authors have any conflict of interest to disclose.
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 Social media: www.elsevier.com/locate/radcr

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https://doi.org/10.1016/j.radcr.2022.10.104
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Case report

A 45-year-old male with a significant history of NF-1 since adolescence presented to his local hospital due to pain and weakness in his back and left lower extremity (LLE), as well as progressive hip flexion weakness. The patient had a tumor resection from his right forearm at 13 years old and a soft tissue lesion from his back at age 31, both reported to have been neurofibromas. At his recent hospitalization, a lumbar spine magnetic resonance imaging (MRI) scan revealed a paraspinal mass concerning for neurofibroma near the L4 nerve root, involving the patient’s left psoas muscle and extending into the neural foramen. After a subsequent tumor debulking surgery, final pathology revealed a low-grade MPNST. A follow-up computed tomography angiogram (CTA) of the abdomen and pelvis with and without intravenous (IV) contrast and 3-D post-processing demonstrated a soft tissue mass arising from the left T12-L1 neuroforaminal canal, measuring approximately 5.8 cm, with mass effect on the left hemidiaphragm, lytic erosion of the L1 left transverse process, and widening of the canal. Additionally, an enhancing left paravertebral mass, measuring approximately 6.1 cm, extending into the left psoas muscle and eroding into the L4 vertebral body and left pedicle was also visualized (Fig. 1). Consequently, the patient was taken to surgery for a complex resection of the mass followed by reconstructive spine surgery. Ultimately, pathological analysis of the lesion revealed a high-grade 6.5 cm MPNST.

Discussion

This article reviews a case of MPNST, a nerve sheath tumor that emerges from either a peripheral nerve, an existing peripheral nerve sheath tumor, or in the context of NF-1 [5]. NF-1, considered the most common hereditary cancer predisposition syndrome, is an autosomal-dominant genetic disorder that arises through the loss-of-function mutation of the NF1 gene. This consists of roles such as microtubule binding, regulation of cyclic AMP levels, and RAS GTTPase-activating protein activity, which is believed to be the main contributor of NF-1-related neoplasia [6,9–11]. More specifically, NF1 stimulates the hydrolysis of Ras-bound GDP to GTP, hence inactivating Ras. NF1 mutations result in hyperactive Ras signaling, triggering abnormal cell proliferation [6,12]. Approximately 8%–13% of all individuals with NF-1 develop MPNSTs [13].

We report a 45-year-old male with an extensive history of NF-1 who presented with back and LLE pain as well as weakness. Subsequent imaging and surgical pathology revealed a significant MPNST, extending from the neural foramen to the psoas muscle. MPNST, when associated with NF-1, is more frequently found in men with a mean age of 28, compared to sporadic MPNST, which occurs equally among men and women with a median age of 41 [4,14,15]. Commonly arising in the proximal portions of the upper and lower extremities, typical sites of involvement comprise nerve roots and bundles in the pelvis and extremities [15]. Patients often exhibit rapidly enlarging masses that frequently induce prompt pain, weakness, and paresthesia [16]. Furthermore, worsening or new-onset pain in the neurofibromas of NF-1 patients should be a cause for concern and evaluation for MPNST [4].

Currently, clinical management for high-grade MPNST in...
cal recurrence (occurring in 40%-65% of patients), metastasis (found in 30%-60% of patients), and unresectable tumors yield an unfavorable prognosis and low survival rates [1,17–20]. Additionally, patients with high-grade MPNSTs have an estimated 5-year survival rate of 20%-50%, and a mortality rate of approximately 75% [1,2,21]. Consequently, early detection and intervention is critical for patient survival.

Computed tomography (CT) imaging may be utilized to determine the size, invasiveness, and relationship with adjacent structures of an MPNST, as well as the presence of distant metastatic disease [1,22]. MPNSTs may calcify, hemorrhage, or erode into adjacent osseous structures, and may show peripheral or irregular enhancement in both CT and MRI [23]. MRI has long been the preferred imaging technique for MPNSTs, since it provides abundant information regarding tumor characterization and the topographical relationship with neighboring structures. However, cinematic rendering (CR) 3-D post-processing serves as a complementary tool to CT enhancing the value of multidetector CT in evaluation and treatment planning of MPNSTs. This reconstruction method provides a remarkable amount of anatomic detail with photorealistic display of skeletal structures, as well as enhanced vessels and soft tissues. It depicts clinically important aspects of MPNSTs and aids in patient workup and surgical planning. Given the novelty of cinematic rendering, its application in detecting MPNSTs, defining clinically significant aspects of these lesions, and assisting in surgical planning has not yet been determined [24].

Fig. 1 – Continued
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