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Case Report

Malignant Peripheral Nerve Sheath Tumor Manifesting as Severe Buttock Pain

Şiddetli Kalça Ağrısı ile Ortaya Çıkan Malign Periferik Sinir Kılıfı Tümörü

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ABSTRACT

A 28-year-old woman presented with complaints of severe buttock pain exacerbating for 2 weeks. Physical examination found numerous café-au-lait macules and axillary freckles. Neurological examination revealed sensory loss at the S3-S5 dermatome and significant vesicorectal dysfunction. Magnetic resonance imaging revealed an enhanced intraspinal mass at S1-S2, with bony erosion on the dorsal aspect of the sacrum. Intraoperatively, the rostral part of the tumor was found to involve a filament of the cauda equina. The tumor protruded extraspinally through an irregular-shaped defect in the dorsal dura and bony erosion. Total resection was achieved. The histological appearance was consistent with malignant peripheral nerve sheath tumor and loss of neurofibromin. Malignant peripheral nerve sheath tumor should be included in the differential diagnosis of spinal tumor in a patient with neurofibromatosis type 1.

KEYWORDS: Malignant peripheral nerve sheath tumor, Dysuria, Neurofibromatosis type 1

ÖZ

Yirmisekiz yaşında kadın hasta son 2 haftadır şiddetlenen ciddi kalça ağrısı şikayeti ile başvurdu. Fizik muayenesinde pekçok café-au-lait lekeleri ve aksiller çiller tesbit edildi. Nörolojik muayenesinde S3-S5 dermatomunda duyu kaybı ve belirgin vezikorektal disfonksiyon saptandı. Manyetik rezonans görüntüleme S1-S2 seviyesinde kontrast tutan intraspinal kitle lezyonu ve sakrumun dorsal yüzünde kemik erozyonu olduğunu gösterdi. İntraoperatif olarak tümörün rostral kısmının kauda ekuina liflerini invaze ettiği görüldü. Tümör ekstraspinal olarak dorsal duradaki irregüler bir defektten dışarı doğru protrude olmaktaydı ve kemik erozyonu yapmaktaydı. Total rezeksiyon yapıldı. Histolojik tanı malign periferik sinir kılıfı tümörü ve nörofibromin kaybı idi. Malign periferik sinir kılıfı tümörü, nörofibromatozis tip 1 tanısı olan ve spinal tümörü olan hastaların ayırıcı tanısında akılda bulundurulmalıdır.

ANAHTAR SÖZCÜKLER: Malign periferik sinir kılıfı tümörü, Disüri, Nörofibromatozis tip 1

INTRODUCTION

Spinal neurinomas are most frequently found in the lumbosacral region (2). In contrast, neurofibroma rarely occurs in this region (8). Neurofibroma may transform into malignant peripheral nerve sheath tumor (MPNST), a rare sarcoma occasionally found in association with neurofibromatosis type 1 (NF1) (6). MPNST accounts for 12% of soft tissue sarcomas and 0.65% of nerve sheath tumors originating in the central nervous system (6,9). High-grade MPNSTs, in particular, have aggressive behavior with dismal prognosis (4). Histological diagnosis and grading of MPNSTs, however, have not been defined due to inconsistent documentations (4,12). Recently, immunohistochemical staining for S-100 protein and neurofibromin were proposed as useful indicators as MPNST (10,12). Total resection is the optimum treatment and the most reliable prognostic factor (1, 5, 9, 11). Here we report a case of MPNST associated with NF1 manifesting as severe buttock pain.

CASE REPORT

An otherwise healthy 28-year-old woman presented with severe buttock pain exacerbating for 2 weeks. Physical examination found numerous café-au-lait macules and axillary freckles. Neurological examination noted sensory loss at the S3 to S5 dermatome, urinary retention, and anal sphincter dysfunction. Computed tomography (CT) scans revealed bony erosion on the dorsal aspect of the S1-S2 (Figure 1A). Magnetic resonance (MR) imaging of the lumbar spine confirmed an intraspinal oval mass 33x16x11 mm in diameter, appearing as isointense on both T1- and T2-weighted images with heterogeneous enhancement (Figure 1B, C). MR imaging of the cerebral and cervicothoracic spine did not identify any pathological conditions. The patient underwent tumor resection through hemilaminectomy of the L5 and dorsal sacral osteotomy. The tumor was found to extend extraspinally, through an irregularly-shaped defect in the dorsal dura and bony erosion (Figure 2). Further, the rostral

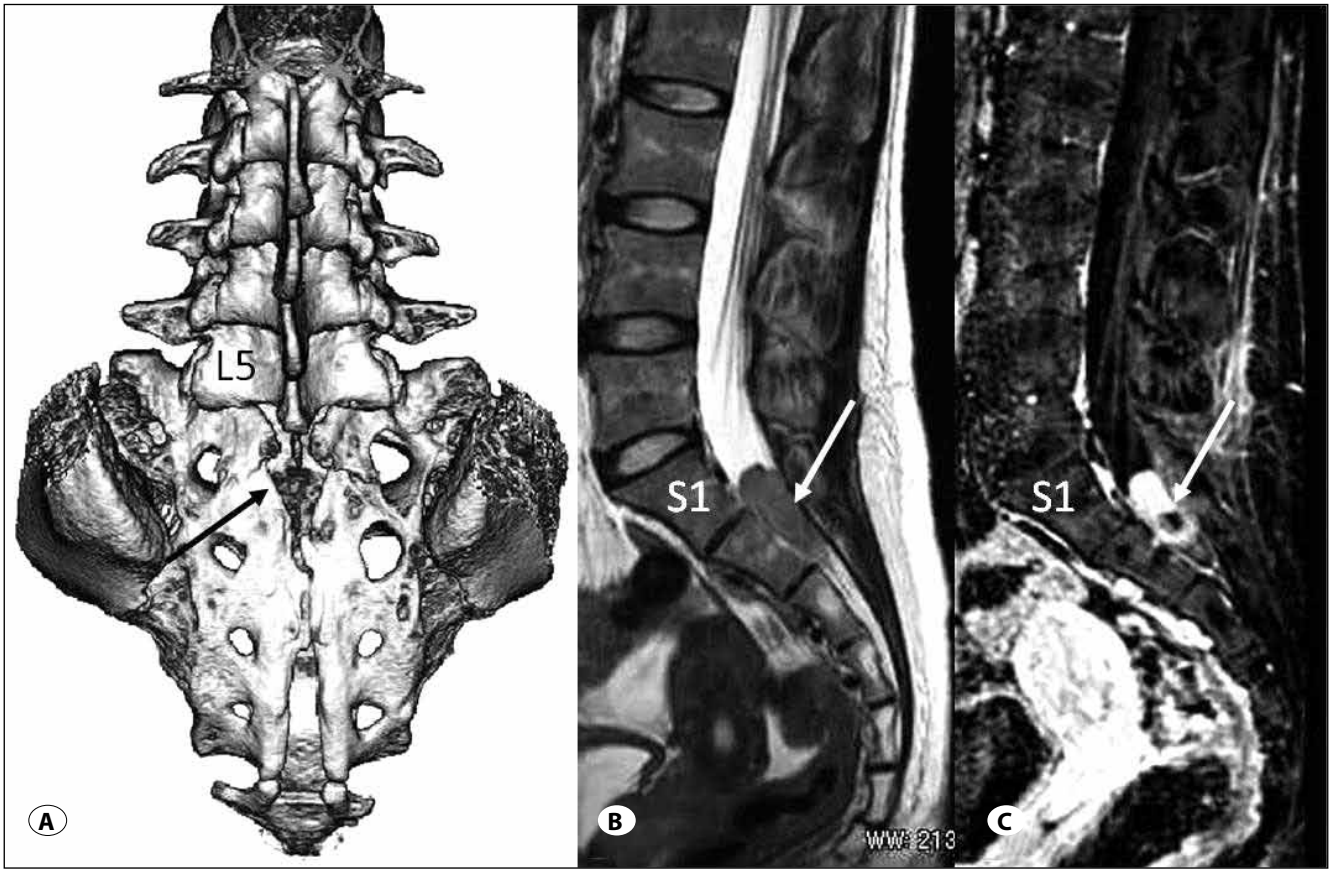

Figure 1: (A) Rear view of three-dimensional computed tomography scans of the sacrum showing a bony defect on the dorsal aspect of the S1-S2 (arrow). Sagittal T2-weighted **(B)** and T1-weighted with contrast medium **(C)** MR images demonstrating an isointense, oval tumor at S1-S2 with heterogeneous enhancement (**B, C:** arrow).

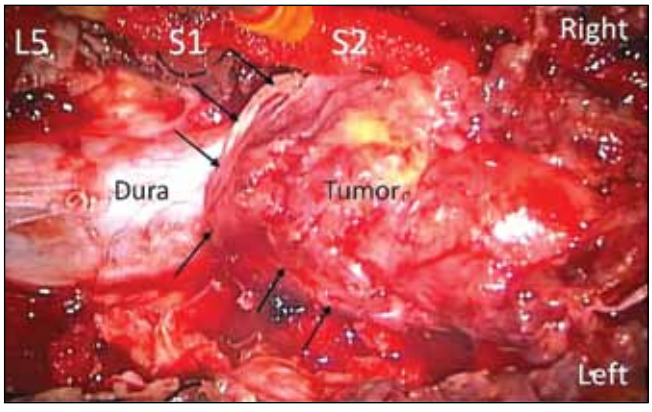

Figure 2: Intraoperative photograph demonstrating the tumor protruding extradurally through an irregularly-shaped defect in the dorsal dura mater (arrows).

part of the tumor involved a filament of the cauda equina. Total resection was achieved. Histological examination showed storiform proliferation of atypical spindle-shaped cells. No nuclear pleomorphism or microvascular proliferation was noted with infrequent mitotic figures of 1-2 per 10 high power fields. Immunohistochemical staining was positive for S-100 protein and vimentin, while negative for epithelial membrane antigen and neurofibromin. The MIB-1 index was 15% in the highest areas. These findings were consistent with NF1-associated MPNST (Figure 3A-F). The patient's buttock pain improved postoperatively, while sensory and vesicorectal dysfunction persisted. MR imaging confirmed total removal of the tumor (Figure 4). Whole-body positron emission tomography (PET)/CT performed postoperatively did not identify any high accumulation area.

DISCUSSION

The majority of neurogenic tumors originating from the sacral region are neurinomas, whereas neurofibroma and MPNST are rare (8). In a large series of spinal neurinoma, NF1 was found in 6.9% (2), whereas MPNST was associated with NF1 in 21% (6). Ren et al. (9) found that one of the 5 MPNSTs of the cauda equina was complicated by NF1.

The histological grade of MPNST varies from II to IV based on the cellularity, nuclear atypia, mitotic index, pleomorphism, microvascular proliferation, and necrosis. However, descriptions of previous cases were inconsistent (4,12). In the present case, the histological findings showed low-grade appearance, whereas the large dural defect and bony erosion suggested

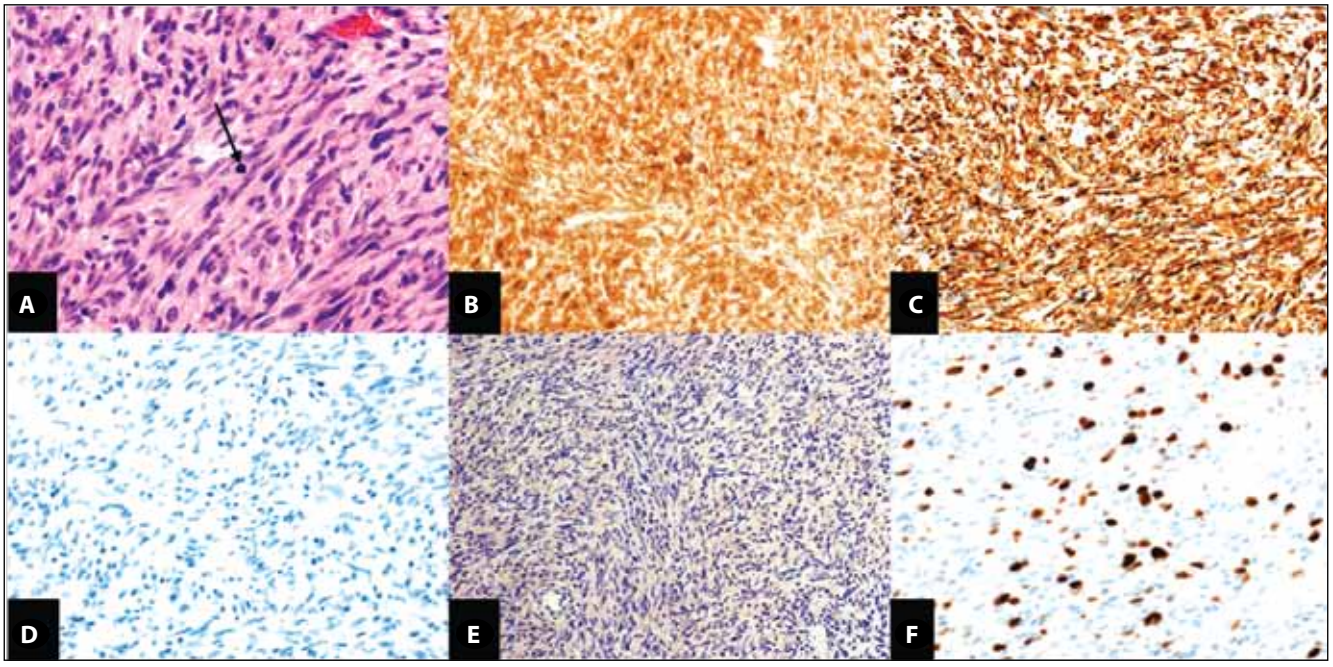

Figure 3: Photomicrographs of the resected specimen. **A)** Hematoxylin and eosin stain showing proliferation of atypical spindle-shaped cells lacking nuclear pleomorphism or microvascular proliferation, with infrequent mitotic figures, which are arranged in a storiform pattern (**a**, arrow). **B-E)** Immunohistochemical examination demonstrating positive staining for S-100 protein **(B)** and vimentin **(C)**, but negative staining for epithelial membrane antigen **(D)** and neurofibromin **(E)**. **F)** MIB-1 index was 15% in the highest areas. Original magnification: **A-D** and **F**, x400; **E**, x200.

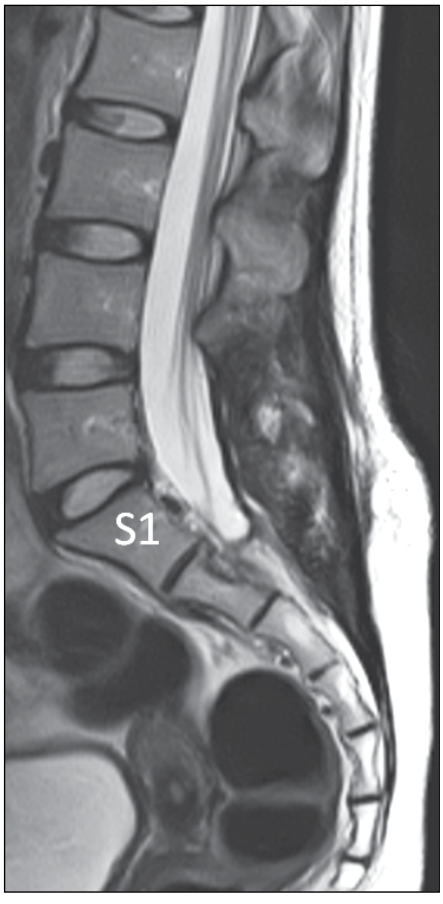

Figure 4: Sagittal T2-weighted MR image performed postoperatively showing total removal of the tumor.

aggressive behavior of the tumor. Four of the previous 5 MPNSTs of the cauda equina were low grade (9).

PET/CT can distinguish MPNST and benign peripheral nerve sheath tumor, in addition to high sensitivity for MPNST, and is useful for both staging and planning treatment strategy (3,7). Radiotherapy exceeding 60 Gy may be an effective management (11), but no adjuvant therapy has been established for MPNST. Therefore, to date, total resection is the key for preferable treatment outcome (1, 5, 9, 11). A previous study did not find NF1 as a prognostic factor (6).

In the present study, severe buttock pain resolved postoperatively. We assumed that cord tethering by the tumor was released at surgery, which might have contributed to the improvement of the pain.

MPNST should be included in the differential diagnosis of spinal tumor in a patient with NF1.

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