Primitive Neuroectodermal Tumour: A Rare Association with Neurofibromatosis Type 1

Hatice Gümüş1, Metehan Gümüş2, Uğur Fırat3, Hakan Önder4, Fatih İnci5
1Department of Radiology, 2Department of General Surgery, 3Department of Pathology, 4Department of Radiology, Medical School, Dicle University, Diyarbakir, 5Department of Radiology, Elazığ Harput State Hospital, Elazığ, Turkey

Abstract
Neurofibromatosis type 1 (NF–1), is a common hereditary disease which often take the form of benign tumours. Spinal primitive neuroectodermal tumour (PNET) is very rare. Herein we report US, CT and MRI findings of a female patient with PNET involving the spinal epidural and paravertebral spaces with neurofibromatosis type 1. A 37-year-old woman has NF–1 presented with lower back pain and progressive weakness of the right lower extremities. US revealed a heterogenous well defined solid mass. CT and MRI revealed heterogeneously enhanced mass originating from the right neural foramen at the level of L5-S1. Adjacent to the lesion, there was a large hemia sac, including bowel loops and mesentery, in the posterolateral abdominal wall. The definitive diagnosis was made by transabdominal tru-cut biopsy. Although it is rare, the possibility of PNET should be kept in mind in assessing the spinal epidural lesions.

Keywords
Spinal Tumour; Primitive Neuroectodermal Tumour; Neurofibromatosis Type 1
Introduction
Neurofibromatosis type 1 (NF–1), is a common hereditary disease in which the skin, nervous system, bones, endocrine glands, and sometimes other organs are the sites of a variety of congenital abnormalities, which often take the form of benign tumors [1]. Spinal primitive neuroectodermal tumor (PNET) is very rare. Thirty-one patients have been reported with such tumors [2], and most of the tumors were intramedullary; only 7 patients had tumors located epidurally. PNETs are aggressive neoplasias that usually diagnosed in infancy. PNET in adulthood is rare and, exceptionally, in association with neurofibromatosis type 1 (NF–1) [3]. Bohn Sarmiento et al. [3] reported a patient with neurofibromatosis type 1 who had lumbar region intra–spinal PNET. To our knowledge there are a few cases of PNET combined with neurofibromatosis type 1 in the English literature [3, 4, 5].

Case Report
A 37-year-old woman presented with lower back pain projecting down to the right leg with progressive weakness of the right lower extremities. The patient with multiple cafe-au-lait spots and neurofibromas in the skin had been diagnosed neurofibromatosis type 1 previously. Transabdominal US revealed a heterogeneous well defined solid mass with central necrosis displacing iliac veins laterally in the right lower quadrant. In the right posterolateral adjacent to the mass, there was a large hernia sac including bowel loops and mesenteric vascular structures (Fig. 1).

CT revealed bilateral meningocele at the level of L5–S1. At this level, a heterogeneous necrotic centered mass with well-defined borders was originating from the right neural foramen. The large, hypodense, heterogenously enhanced mass caused vertebral scalloping, and displaced bowels and vascular structures laterally. Adjacent to the lesion, there was a large hernia sac, including bowel loops and mesentery, in the posterolateral abdominal wall (Fig. 2). The mass was hipointense on T1W, hipointense on T2W, and enhanced heterogenously on MRI (Fig. 3). Cranial MRI was normal.

Discussion
Ewing's sarcoma /PNET arising from the lumbosacral spine is reported very rarely. PNET needs to be differentiated from other tumors ( schwannoma, neurofibroma, lymphoma, malignant peripheral nevre sheath tumors (MPNST) ) which are arising in
this region [6]. The most useful diagnostic imaging methods are computerized tomography (CT) and magnetic resonance imaging (MRI) [7]. PNET may exhibit a well-defined hypodense mass displacing the thecal sac on nonenhanced CT scans, and heterogeneous contrast enhancement on contrast-enhanced CT scans. Central, nonenhancing areas show necrosis within the tumor. MRI is useful for determining the extent of the tumor because of its high soft tissue resolution. The tumor is usually hypo-isointense to the spinal cord on T1-W images and hyperintense on T2-W images. Mild to moderate and homogeneous or heterogeneous enhancement can be seen on postcontrast MRI [7]. Schwannoma is the most common intradural extramedullary neurogenic tumor in the spine. Schwannomas are well-encapsulated, compressing the cord, conus to the contralateral side. These tumors have a cystic component (40% cases), and show peripheral enhancement on postcontrast T1-W images [8]. Neurofibromas are well-defined lesions. The tumors show a hyperintens rim and a hypointense center on T2-W and postcontrast T1-W images. They are also often associated with NF–1 [6]. Schwannoma and neurofibroma were considered in our patient because of CT and MRI characteristics of lesions. Lymphomas (non-Hodgkin’s lymphoma) present in older age-groups and spread via the subarachnoid space, causing diffuse thickening of the spinal nerve roots. They show marked enhancement on postcontrast MRI images [6]. Lymphoma was not considered in our patient because she was 37 year old. Approximately 5% of patients with NF–1 develop MPNST, which usually arises from a plexiform neurofibroma [9]. MPNSTs are usually infiltrative and heterogenous, and have irregular margins on CT and MRI. These tumours have central hemorrhage and necrosis. They show variable patterns of enhancement, and they commonly show diffuse or peripheral heterogeneous contrast enhancement on postcontrast CT and MRI [10]. MPNST was not considered in our patient because CT and MRI showed well-defined lesions. In this case, we established our radiological diagnosis as benign nerve sheet tumor (schwannoma, neurofibroma) in the patient with NF–1. Because of the rare occurrence, spinal PNET was not considered before the histologic examination. Diagnosis could be established only after the biopsy. Differentiation of these rare lesions requires imaging studies along with histopathological confirmation [6]. Spinal epidural PNET is a rare neoplasm that difficult to diagnose. PNET in association with NF–1 is very rare (there is only one other case in literature). CT and MRI seem to be effective in the overall assessment. Although it is rare, the possibility of PNET should be kept in mind in assessing the spinal epidural lesions.

Competing interests

The authors declare that they have no competing interests.

References

3. Bohn SU, Aguiar BD, Camacho GR, Rivero VJC, Aguiar MJ. Lumbar region intraspinal primitive neuroectodermal tumour (PNET) combined with neurofibromato-