LETTERS

Aggressive malignant peripheral nerve sheath tumor of the lower limb in a patient with neurofibromatosis-1 and multiple spinal cord neurofibromas

Dear Editor,

Neurofibromas are nerve sheath tumors, usually benign, of the peripheral nervous system occurring sporadically or in the context of Neurofibromatosis-1 (NFT-1), the most common neurocutaneous disorder (incidence: ~1/3000). Around 20% of NFT-1 patients will develop a plexiform neurofibroma but only 1-2% one will develop a malignant peripheral nerve sheath tumor (MPNST). No reliable screening test exists in clinical practice yet, although recently potential markers for early diagnosis have been researched.

We report an aggressive, large, MPNST of the left thigh in a 29-years-old male patient with progressive cervical myelopathy. The patient presented with tetra-spasticity, gait disturbance and instability. He also suffered from intense pain in the proximal left lower limb due to a large tumor of the medial thigh (8.8 cm in diameter), diagnosed as a plexiform neurofibroma by magnetic resonance imaging (MRI) (Figure 1). The patient had a typical clinical presentation of NFT-1 with multiple asymptomatic cutaneous, subcutaneous and spinal neurofibromas.

He was treated for cervical myelopathy with C2-C5 laminectomy and decompression by removal of intradural tumors and also had the thigh tumor removed en-block. Intraoperative, the thigh tumor was recognized to be larger in dimensions (11cm-diameter) than in the 2-months-old pre-op MRI. Histopathology showed a grade III-IV MPNST (CD56++, focal CD34++, and focal S100+) for the lower limb tumor, while cervical tumors were identified as typical neurofibromas. The patient was referred to an Oncological Department for further treatment but had a local recurrence within 3 months of the original operation. He was treated with excision of the recurrent lesion and also had radiotherapy sessions. He remained free of recurrence at follow-up examination (4 months from the second operation).

NFT-1 and tumor volume have been widely identified as poor independent prognostic factors regarding tumor recurrence and patient survival. Recent meta-analysis, on the other hand, has shown significant improvement of survival for NFT-1 patients with MPNST in the last ten years[1]. In conclusion, any new neurological symptoms in patients with plexifom neurofibromas, as well as new painful swellings in NFT-1 patients should always raise awareness for a possible MPNST[2]. Regular clinical and follow-up examination should not be omitted and treatment should not differ in such patients than in those cases of sporadic tumors. Neural lesions should be surgically treated when they are focal, symptomatic and resectable[3], while biopsy under real-time multimodal imaging an increase in accurate detection of malignancies. Positron Emission Tomography (PET) and PET-CT in particular have had their value demonstrated regarding diagnostic accuracy in NFT-1 associated MPNSTs[3].

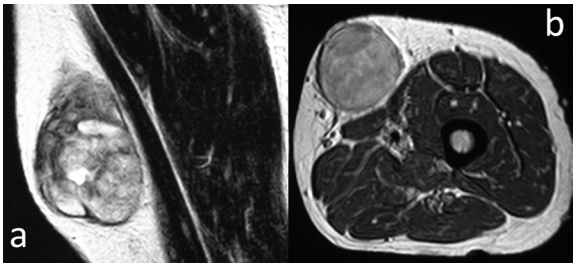

Figure 1: T1-weighted Magnetic Resonance Imaging of the large Malignant Peripheral Nerve Sheath Tumor of the medial thigh (5.65 x 5.25 x 8.8 cm) adjacent to, but not invading, medial vastus and sartorius muscles, a) sagittal plane and b) axial plane at greatest dimensions.

Conflict of Interest

None.

Keywords: Neurofibromatosis-1, malignant peripheral nerve sheath tumor, recurrence, spinal neurofibroma

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