A Huge Malignant Peripheral Nerve Sheath Tumor Revealing Von Recklinghausen’s Disease

Choukri Elm’hadi1,*, Mohammed Reda Khmamouche1, Mehdi Toreis1, Rachid Tanz1, Tarik Mahfoud1, Hassan Errihani2 and Mohammed Ichou1

1Medical Oncology Department, Mohammed V Military Teaching Hospital of Rabat, Morocco
2Medical Oncology Department, National Institute of Oncology Sidi Mohamed Ben Abdellah, Rabat, Morocco

Abstract

Only a few cases of malignant peripheral nerve sheath tumor (MPNST) associated with Von Recklinghausen’s disease or type I neurofibromatosis (NF-1) have so far been reported worldwide. We present a case of a 27 year old man with MPNST of the left thigh associated with NF-1. The diagnosis was based on clinical, radiological and histopathological evidence. He presented a large mass of thigh, deeply adhering, with the presence of collateral venous circulation. He also presented multiple café-au-lait spots, with a many neurofibromas. MRI of the hip and left thigh showed the presence of a bulky tissue process, badly limited, measuring 24.6×11×12 cm occupying the anterolateral and posterolateral lodge with an intermediate signal in T1, discreetly more intense in T2. The microscopic and immunohistochemical findings supported the final diagnosis of MPNST with mesenchymal differentiation. The staging was negative. Also, the diagnosis of NF-1 is held according to the presence of two NIH criteria. The decision of the multidisciplinary meeting was to make a neoadjuvant chemotherapy to surgery with a doublet of adriamycin and ifosfamide with surveillance for other tumor development or multisystem complications. The presence of a large mass on the path of a peripheral nerve requires a careful examination of the skin for signs evoking a von Recklinghausen disease.

Keywords: Malignant peripheral nerve sheath tumor; Neurofibromatosis; Von Recklinghausen’s disease

Abbreviations: NF1: Neurofibromatosis 1; MPNST: Malignant Peripheral Nerve Sheath Tumor; NIH: National Institutes of Health; EMA: Epithelial Membrane Antigen; MRI: Magnetic Resonance Imaging; CT: Computer Tomography; WHO: World Health Organization

Introduction

Neurofibromatosis 1 (NF1) or von Recklinghausen’s disease is a genetic disease characterized by a high variability of clinical expression. Diagnosis is usually clinical. Malignant transformation is rare and dreadful. We present a case revealed by a huge malignant peripheral nerve heath tumor revealing von Recklinghausen’s disease.

Case Report

We present a case of a Moroccan 27 year old men, with no particular history which reports the appearance a mass of the upper and outer of the left thigh gradually increasing volume. No family or personal histories of neurological, musculoskeletal, dermatological or visual disease were noted. On medical examination, the patient has café au lait spots in different sizes and shapes on all over the body accentuated in the trunk (Figure 1) with the presence of many neurofibromas in the dorsal trunk and roots of members. The mass of thigh was large, hard, deeply adhering, with the presence of collateral venous circulation (Figure 2). Blood pressure was 110/70 mmhg. The patient does not present cognitive impairment, focal neurological deficits or skeletal anomalies and visual acuity and fundus were normal. A biopsy of the mass of thigh was performed, histological examination has shown a very dense population of atypical spindle cells with wavy, hyperchromatic nuclei and high mitotic activity with Alternating hypercellular/hypocellular region. The results of immunohistochemical staining showed positivity for S-100 protein, CD-34, EMA, actine muscle lisse ,neurofilament, desmine, myogene and Ki-67 for 30%. The microscopic and immunohistochemical findings supported the final diagnosis of MPNST with mesenchyme differentiation. Meanwhile, The diagnosis of neurofibromatosis type I is held according to the presence of two of National institutes of health (NIH) Consensus Development Conference criteria:1) Six or more café au lait macules larger than 1.5 cm in post pubertal individuals 2)Two or more neurofibromas of any type . MRI of the hip and left thigh showed the presence of a bulky tissue process, badly limited, measuring 24.6×11×12 cm occupying the anterolateral and poster lateral lodge with an intermediate signal in T1, discreetly more intense in T2, encompassing deep vascular pedicle with multiple hypo intense areas in enhancement after gadolinium injection.

Figure 1: Café au lait spots in different sizes and shapes in the trunk.

*Corresponding author: Choukri Elm’hadi, Medical Oncology Department, Mohammed V Military Teaching Hospital of Rabat, Morocco, Tel: 00212613144918; E-mail: dr.choukrielmhadi@hotmail.com

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showing a necrosis. The brain CT and thoraco-abdominal-pelvic showed no secondary location and not any deep neurofibroma. Bone scan showed fixing heterogeneous left femoral shaft. The decision of the multidisciplinary meeting was to make a neoadjuvant chemotherapy to surgery with a doublet of Adriamycin and ifosfamide with surveillance for other tumor development or multisystem complications.

**Discussion**

Malign Peripheral Nerve Sheath Tumors (or MPNST) are defined by the WHO classification in 2013 as developed malignancies [1] from a peripheral nerve; or a benign tumor of the nerve sheaths; or in a patient with neurofibromatosis type 1. These are a very rare tumor with approximately 5-10% of all soft tissue sarcomas [2]. The incidence in the general population is 0.001% and 5-10% of NF1 patients [3] often in a sporadic fashion and rarely radiation-induced. They usually appear between the third and fourth decade but earlier disclosure is particularly possible in NF1 patients [4]. The sex ratio is close to 1 with a slight female predominance [4].

Most MPNSTs develop mainly with peripheral nerves such as the sciatic nerve, the brachial plexus and the sacral plexus. They are usually deep-seated and often involving the proximal upper and lower extremities as well as the trunk [3]. The clinical expression is variable including radicular pain, parenthesis, motor weakness or enlarging palpable mass whose Rapid increasing should raise malignant degeneration of a neurofibroma often in the setting of NF1 [5].

Imaging studies have a dual purpose: to distinguish between benign tumors and MPNST and specify the local and general extension of these tumors. MRI is the imaging modality of choice for characterizing MPNST. These tumors share basic imaging characteristics with their benign counterparts such as neurofibromas and schwannomas they are distinguished by four 4 features: a size ≥ 5 cm; peripheral enhancement; edematous areas périlésionnelles; and cystic areas intratumoral distinguished by four 4 features: a size ≥ 5 cm; peripheral enhancement; edematous areas périlésionnelles; and cystic areas intratumoral.

In all cases, the prognosis is poor with an overall 5-year survival of 25% in case of NF1 and 50% in case of isolated tumor [11]. Local recurrence is frequent and metastases (lung, liver, skin, bone) that appear within an average of 2 years. Patients with NF1 were previously thought to have a worse prognosis than did patients with sporadic MPNSTs [13].

**Conclusion**

The presence of a large mass on the path of a peripheral nerve requires a careful examination of the skin for signs evoking a von Recklinghausen disease.

**Competing Interests**

The authors declare that they have no competing interests.

**References**


