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

Choukri Elm'hadi, Mohammed Reda Khmamouche, Mehdi Toreis, Rachid Tanzi, Tarik Mahfoud, Hassan Errihani and Mohammed Ichou

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 dorsum and root 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 posterolateral 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

Received April 02, 2016; Accepted May 18, 2016; Published May 27, 2016

Citation: Elm’hadi C, Khmamouche MR, Toreis M, Tanzi R, Mahfoud T, et al. (2016) A Huge Malignant Peripheral Nerve Sheath Tumor Revealing Von Recklinghausen’s Disease. J Integr Oncol 5: 170. doi: 10.4172/2329-6771.1000170

Copyright: © 2016 Elm’hadi C, et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.
The pleura. For this reason, a chest computed tomography scan is the preferred imaging study to screen for distant disease. A bone scan should also be obtained to help identify metastatic bone disease.

**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, paresthesia, 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 MPNSTs and specify the local and general extension of these tumors other differential diagnoses (melanoma, fibrosarcoma, synovial sarcoma monophasic, leiomyosarcoma or more rarely neurofibroma and schwannoma cell). The most frequently used markers are S-100 protein, Leu-7 and myelin basic protein. S-100 protein is the most widely used antigen for neural differentiation and can be identified in 50%. Leu-7 and myelin basic protein are found in about 50% and 40% of MPNSTs respectively [7]. Ki-67 greater than 20 has identified as being an independent prognostic factor [8].

**Conclusion**

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

**Competing Interests**

The authors declare that they have no competing interests.

**References**

1. Le Guellec S (2015) Nerve sheath tumours. Ann Pathol 35: 54-70.
2. Scheithauer BW, Woodruff JM, Erlandson RA (1999) Primary malignant tumors of peripheral nerve Tumors of the peripheral nervous system. Washington, DC: Amer Registry of Pathology.
3. Beer TC (2012 ) Malignant Peripheral Nerve Sheath Tumor (MPNST): an overview with emphasis on pathology imaging and management strategies.
4. Bouvier C, de Paula AM, Roche PH, Chagnaud C, Figarella-Branger D (2013)
Tumeurs du système nerveux périphérique. EMC-Neurologie 10: 1-11.

5. Ferber RE, Gutmann DH (2002) International consensus statement on malignant peripheral nerve sheath tumors in neurofibromatosis. Cancer research 62: 1573-1577.

6. Wasa J, Nishida Y, Tsukushi S, Shido Y, Sugiura H, et al. (2010) MRI features in the differentiation of malignant peripheral nerve sheath tumors and neurofibromas. AJR Am J Roentgenol 194: 1568-1574.

7. Enzinger FM, Weiss SW (1995) Malignant tumors of the peripheral nerves. In: Soft Tissue Tumors. 3rd edition pp. 889-928.

8. Heslin MJ, Cordon-Cardo C, Lewis JJ, Woodruff JM, Brennan MF (1998) Ki-67 detected by MIB-1 predicts distant metastasis and tumor mortality in primary, high grade extremity soft tissue sarcoma. Cancer 83: 490-497.

9. Savar A, Cestari DM (2008) Neurofibromatosis type I: genetics and clinical manifestations. Semin Ophthalmol 23: 45-51.

10. DeBella K, Szudek J, Friedman JM (2000) Use of the national institutes of health criteria for diagnosis of neurofibromatosis 1 in children. Pediatrics 105: 608-614.

11. Charfeddine I, Mnejja M, Hammami B, Chakroun AM, Kallel S, et al. (2008) Tumeur maligne des gaines nerveuses périphériques révélant une neurofibromatose type 1. Journal Tunisien d’ORL et de Chirurgie Cervico-Faciale 20: 1.

12. Riccardi VM (1992) Neurofibromatosis: Phenotype, Natural History and Pathogenesis. 2nd edition.

13. Cashen DV, Parisien RC, Raskin K, Hornicek FJ, Gebhardt MC, et al. (2004) Survival data for patients with malignant schwannoma. Clin Orthop Relat Res pp: 69-73.