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CASE REPORT

Giant malignant peripheral sciatic nerve sheath tumour causing paralysis in a patient with neurofibromatosis 1

D. Sáez Martínez[a,*], A. Caballero[b], J.A. Calvo[a], M. Cuervo[c] and J. Vaquero Martín[a]

[a] *Servicio de COT, Hospital General Universitario Gregorio Marañón, Madrid, Spain*
[b] *Servicio de COT, Hospital de Fraternidad Muprespa, Madrid, Spain*
[c] *Servicio de Oncología Musculoesquelética, Hospital General Universitario Gregorio Marañón, Madrid, Spain*

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KEYWORDS
Neurofibroma;
Neurofibromatosis;
Peroneal nerve paralysis

Abstract
Objective: A malignant peripheral nerve sheath tumour is a sarcoma arising from nerve tissue.
Material and method: We present the case of a giant buttock tumour in an NFM 1 patient causing peroneal nerve paralysis.
Results: In up to 50%of malignant peripheral nerve sheath tumours (MPNST) an association with neurofibromatosis 1 (NFM 1) is found.
Discussion and conclusions: In patients with NFM 1, a sudden increase in size of a previously stable neurofibroma should be viewed with great suspicion of malignant transformation and lead to an immediate biopsy

PALABRAS CLAVE
Neurofibroma;
Neurofibromatosis;
Parálisis nervio ciático

Tumor gigante maligno dependiente de la vaina del nervio ciático como causa de parálisis en paciente con neurofibromatosis 1

Resumen
Objetivo: Los tumores malignos dependientes de la vaina de un nervio periférico son una variedad de sarcomas que demuestran diferenciación tisular nerviosa.
Material y método: Presentamos el caso de una tumoración glútea gigante en una paciente afecta de neurofibromatosis tipo 1 como causa de parálisis de CPE.
Resultado: Hasta en un 50% de los casos se asocian a pacientes con neurofibromatosis tipo 1.
Discusión y conclusiones: En un paciente afecto de NFM 1, el aumento de tamaño de un neurofibroma previamente estable debe hacer sospechar la transformación maligna y por lo tanto conllevar una biopsia inmediata.

* Corresponding author.
E-mail: saezmart@yahoo.es (D. Sáez Martínez).

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Introduction

The malignant peripheral nerve sheath tumours (MPNST) are a type of soft tissue sarcoma that exhibit nerve tissue differentiation. Most cases (half of all MPNSTs) are associated with type 1 neurofibromatosis (NFM 1) and up to two thirds appear on prior neurofibromas. We present the case of a female patient with an NFM 1 who consults because an increase in size of a gluteal swelling causing paralysis of the external popliteal sciatic nerve (EPS).

Case report

Forty-nine year old female with a history of type 1 neurofibromatosis who is admitted to our centre with swelling of the left buttock (fig. 1); she reports progressive increase in size of the mass in the last few months, as well as intense sciatica type pain in her lower left leg.

Upon examination, a 10×5 cm swelling is detected; with rock-hard consistency, adhered to deep planes. Likewise, the patient presented paresis of muscles groups dependent on the sciatic nerve with greater involvement of the EPS territory.

The MRI study revealed a soft tissue mass arising from the sciatic nerve measuring 7.98×6.53×8.16 cm entering the greater sciatic foramen (fig. 2). The extension study (CT of the chest, abdomen, and pelvis, and bone tracing) only brought out the presence of a lesion of the mediastinum that was consistent with adenopathy or neurofibroma.

An incisional biopsy was originally performed but yielded inconclusive results. As a result, 3 weeks later the tumour was removed with intra-operative biopsy results confirming malignancy based on its high mitotic index. The decision was therefore made to carry out a wide excision of the specimen with both proximal (roots of the lumbo-sacral plexus) and distal (sciatic nerve) margins (fig. 3). Macroscopically, it was a swelling measuring 10×5 cm arising from the sciatic nerve that extended from the sciatic notch to the distal medial third of the sciatic nerve (fig. 4).

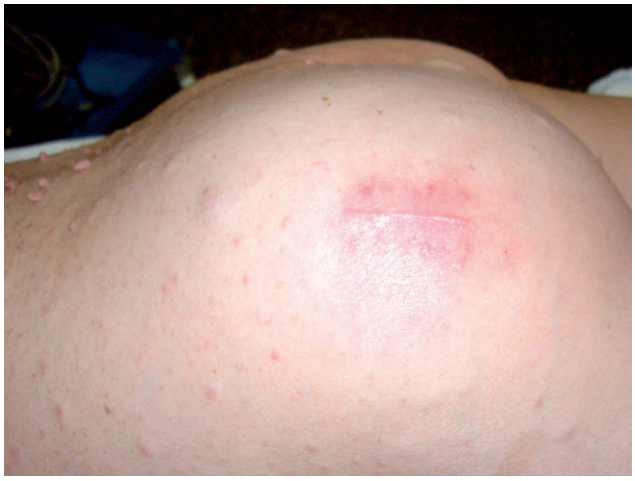

Figure 1 Image of gluteal swelling with neurofibromas.

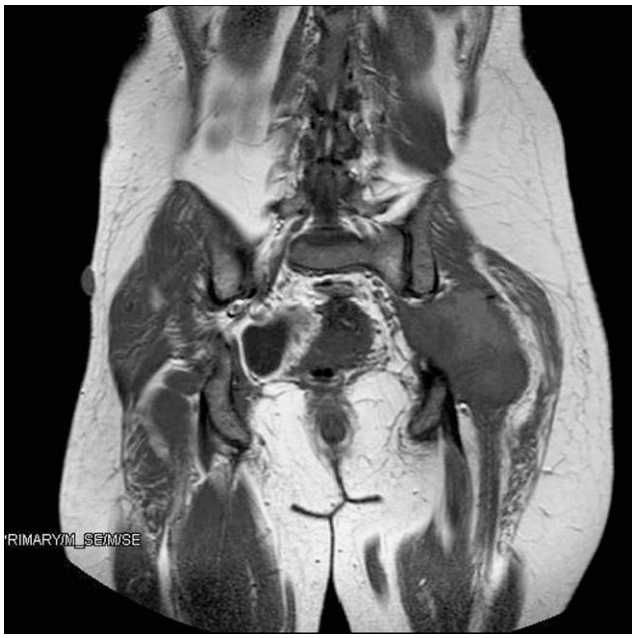

Figure 2 MRI.

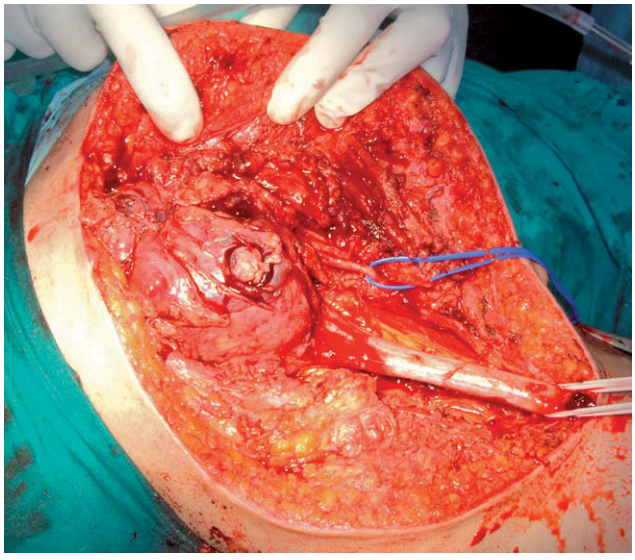

Figure 3 Intra-operative image of the tumour.

The postoperative coursed without incident and the patient was fitted with a drop foot brace.

The histopathological diagnosis confirmed that it was a malignant nerve sheath tumour with markedly necrotic tissue, proliferation of S-100 positive fusiform elements, having a Ki67 cell proliferation index of 20% (fig. 5). The patient was referred to the radiotherapy department for adjuvant radiotherapy.

Discussion

MPNSTs are a type of spindle cell sarcomas arising from a nerve or neurofibroma or that display nerve tissue

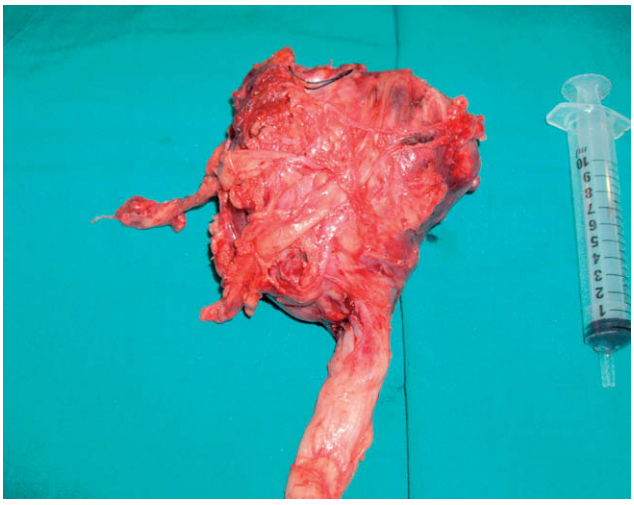

Figure 4 Surgical specimen.

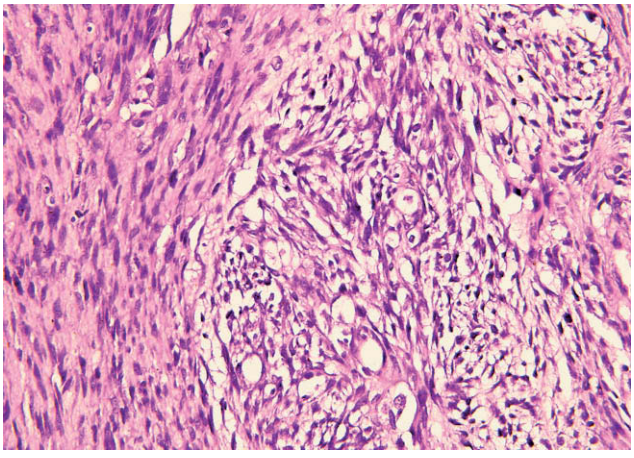

Figure 5 Pathological anatomy.

differentiation. They have also been known as malignant Schwannomas, neurogenic sarcomas, malignant neurilemmomas, or neurofibrosarcomas.

They mainly affect patients aged 20-50 years, accounting for 5-10% of all soft tissue sarcomas. MPNSTs tend to develop at the expense of main nerve trunks including the sciatic nerve, brachial plexus, and sacral plexus. Clinically, they manifest with pain and motor and sensory neurological impairment more often than is the case with benign peripheral nerve sheath tumours.

Most cases are associated with NFM 1 and up to two thirds appear on previously existing neurofibromas. On the one hand, only 5% of the patients with NFM 1 develop MPNST. MPNSTs can also be masses that are secondary to prior radiotherapy, appearing after protracted periods of time (10-20 years).[1] In a patient with NFM 1, enlargement of a previously stable neurofibroma should lead us to suspect malignant transformation and a biopsy should therefore be performed immediately.[2]

Microscopically, MPNSTs resemble one another, since their cells have Schwann cell characteristics, alternating intensely hyper- and hypocellular areas; areas of tumour necrosis are also common.

MPNSTs share the radiographic characteristics of neurogenic tumours, with an elongated shape that runs longitudinally along the distribution of the nerve. Although CT and MRI images rarely aid in identifying malignancy of peripheral nerve tumours, there are some traits that are suspicious; for instance, size larger than 5 cm, heterogeneous content, poorly defined margins, invasion of fatty planes, as well as perilesional oedema.[3,4]

As in the case of the vast majority of soft tissue sarcomas, treatment is based on surgical excision, with radiotherapy as adjuvant treatment.

A high degree of suspicion is necessary when previously silent neurofibromas enlarge in patients with neurofibromatosis 1, principally greater than 5 cm.

References

1. Heiner J, White J. Treatment principles soft tissue tumors. In: Damron TA, editors. Oncology and basic science. 1 ed. Philadelphia: Lippincott Williams and Wilkins; 2008. p. 87-92.
2. Borden ANDC, Baker LH, Bell RS, Bramwell V, Demetri GD, Eisenberg BL, et al. Soft tissue sarcomas of adults: state of the science. Clin Cancer Res. 2003;9:1941-56.
3. Pllavaki M, Chourmouzi D, Kiziridou A, Skordalakai A, Zarampoukas T, Drevelengas A. Imaging of peripheral nerve sheath tumors with pathologic correlation. Eur J Radiol. 2004;52:229-39.
4. Lin J, Martel W. Cross-sectional imaging of peripheral nerve sheath tumors: characteristic signs on CT, MR imaging, and sonography. Am J Roentgenol. 2001;176:75-82.