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Segmental schwannomatosis of the lower limb: About a case

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Abstract
Introduction: Neurofibromatosis and schwannomatosis are a group of related tumour suppressor syndromes characterised by an increased incidence of tumours of the nerve sheath. Schwannomatosis involving the nerves of a single limb is a rare clinicopathologic entity, distinguished from neurofibromatosis type 2 (NF2). The clinical similarities between the two aforementioned conditions may cause diagnostic difficulties. The main diagnostic criterion in favour of NF2 in this case is the presence of bilateral vestibular nerve schwannoma, which is mainly revealed by hearing loss.
Case report: A 41 year-old male underwent surgery three times in 2012, 2019 and 2023 for schwannomas of the fibular nerve, the medial plantar nerve, the saphenous nerve and lastly the sciatic nerve of the right lower limb. Anatomopathological examination confirmed the diagnosis of schwannoma every time. He was relieved of pain and paraesthesia after all his surgeries. The patient was diagnosed with schwannomatosis based on the recurrence of multiple schwannomas of the right lower limb over the course of 11 years and the lack of clinical signs of bilateral hearing loss, tinnitus or balance loss in favour of vestibular tumours characteristic of the NF2.
Conclusion: In cases of multiple schwannomas with no clinical or radiological signs of concomitant bilateral vestibular nerve schwannomas, the diagnosis of schwannomatosis is very likely. It becomes definite after eliminating all criteria for the diagnosis of NF2, including first-degree family history of Von Recklinghausen's disease.

Keywords: Schwannoma, schwannomatosis, neurofibromatosis, surgery

Introduction
Neurofibromatosis and schwannomatosis are a group of related tumour suppressor syndromes characterised by an increased incidence of tumours of the nerve sheath. Segmental schwannomatosis involving the nerves of a single limb or less than five contiguous segments of spine is an uncommon clinicopathologic entity, distinguished from neurofibromatosis type 2 (NF2). The clinical similarities between the two aforementioned conditions may cause diagnostic difficulties.

Case presentation
A 41 year-old Tunisian male with no family or personal history of systemic disease consulted our department in 2012 (at the age of 29 years-old) for chronic paraesthesia of the dorsal aspect of the right foot for the previous two years. On physical examination, he had a palpable, firm, subcutaneous mass under the head of the fibula, measuring 7 by 3 cm. This mass is mobile transversely and fixed longitudinally. The common fibular nerve was evaluated at S2M5 according to the "British Medical Research Council" (BMRC) scale. Tinel's sign was elicited as positive. He had no café-au-lait spots or other signs in favour of a neurofibromatosis (NF) and no family history of NF or schwannomatosis. Sonographic examination showed the presence of a bilobed tumour of the superficial fibular nerve (figure 1). Magnetic resonance imaging (MRI) showed a well-defined, bilobed solid mass of the fibular muscle compartment, measuring 7 x 2.5 cm, with iso-signal in T_1 and heterogeneous hyper-signal in T_2, enhancing heterogeneously after Gadolinium injection. The mass characteristics were very suggestive of a neuroma of the common fibular nerve (figure 2).
Surgical exploration confirmed the presence of two well-defined masses of the superficial fibular nerve with no signs of malignancy or nerve invasion, in favour of a bilobed schwannoma (figure 3). Excisional biopsy of 6*3 cm and 1.5 cm masses was done and histopathology confirmed the diagnosis of schwannoma.

After surgery, the patient recovered a normal sensibility at the dorsal aspect of the right foot with no alteration of the muscles innervated by the superficial fibular nerve (figure 4).

Seven years later, the patient presented with paraesthesia and progressively increasing pain of the right thigh and the plantar aspect of the right foot for the last seven months. Physical examination revealed a palpable firm 4 cm mass of the medial aspect of the thigh and a two cm firm mass of the medial plantar foot and no recurrence of the schwannoma at the operated site. Echography of the lower limb revealed a tumour of the saphenous nerve and another of the medial plantar nerve. Surgery confirmed the presence of a three cm well encapsulated mass of the saphenous nerve situated between the fascicles of the sartorius muscle that was easily extirpated, preserving the nerve fascicles (figure 5). The exploration of the plantar foot revealed a well-defined spheroid mass of the medial plantar nerve measuring 1.6 cm that was enucleated with no difficulty (figure 6). Anatomopathological examination confirmed the diagnosis of schwannoma in both sites.

In 2023, the patient complained of a discomfort of the right gluteal area, exacerbated in the sitting position. A tenderness on palpation and a positive Tinel sign were the only physical examination findings. We noted the presence of 2.5 by 4 cm oval mass of the sciatic nerve at the level of the ischiatic tuberosity. The mass removed surgically was confirmed to be a schwannoma of the sciatic nerve.

The patient was relieved of pain and paraesthesia after all his surgeries.

The patient was diagnosed with schwannomatosis based on the recurrence of multiple schwannomas of the right lower limb over the course of 11 years and the lack of clinical signs of bilateral hearing loss, tinnitus or balance loss in favour of vestibular tumours characteristic of the NF2.

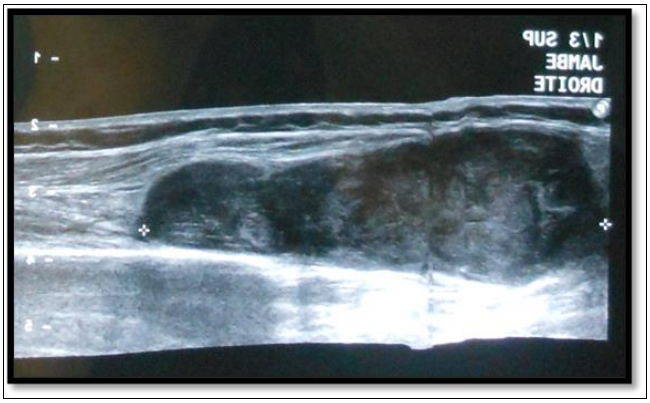

Fig 1: Sonography objectified a homogenous hypoechoic hypovascular mass of the external aspect of the leg, in favour of a nerve tumour.

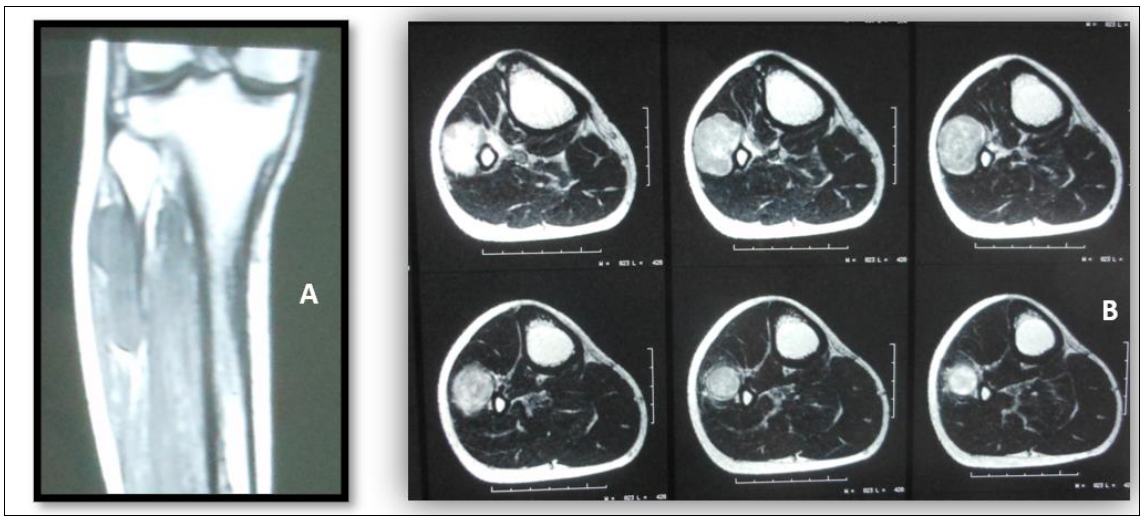

Fig 2: (A) Coronal view of a bilobed mass in the fibular muscle compartment of the leg on T_1-weighted images. (B) Same mass with heterogenous enhancing after gadolinium injection (axial view).

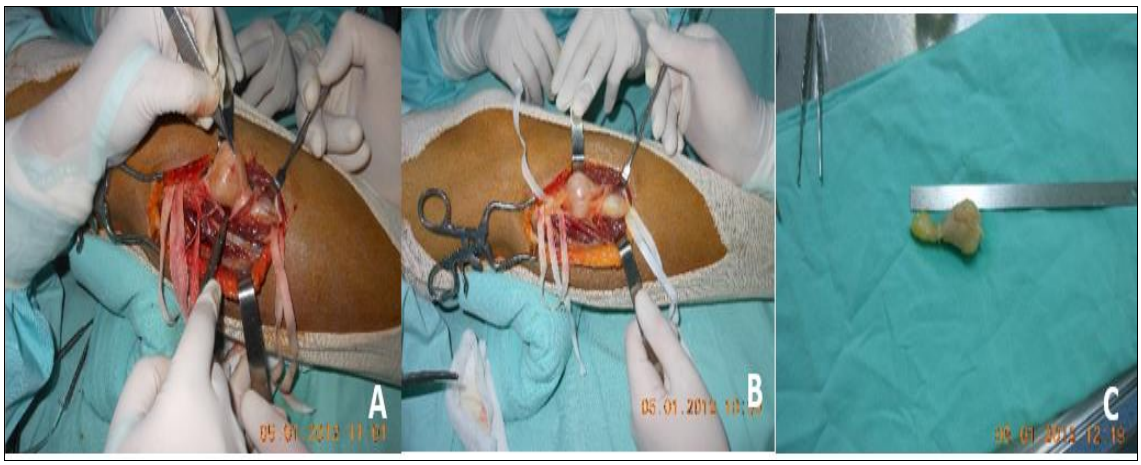

Fig 3: (A-B) Intra-operative image of a well-circumscribed bilobed tumour of the common fibular nerve. (C) Schwannoma tissue completely removed from the fibular nerve.

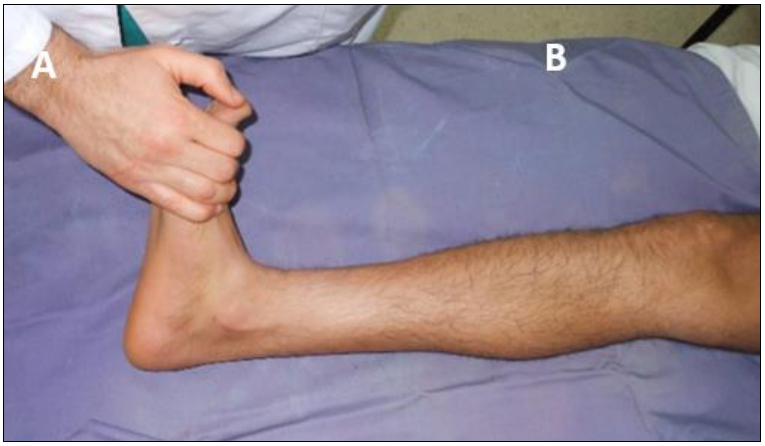

Fig 4: Post-operative evaluation of the operated limb. The patient had no alteration of the motor and sensory function of the common fibular nerve.

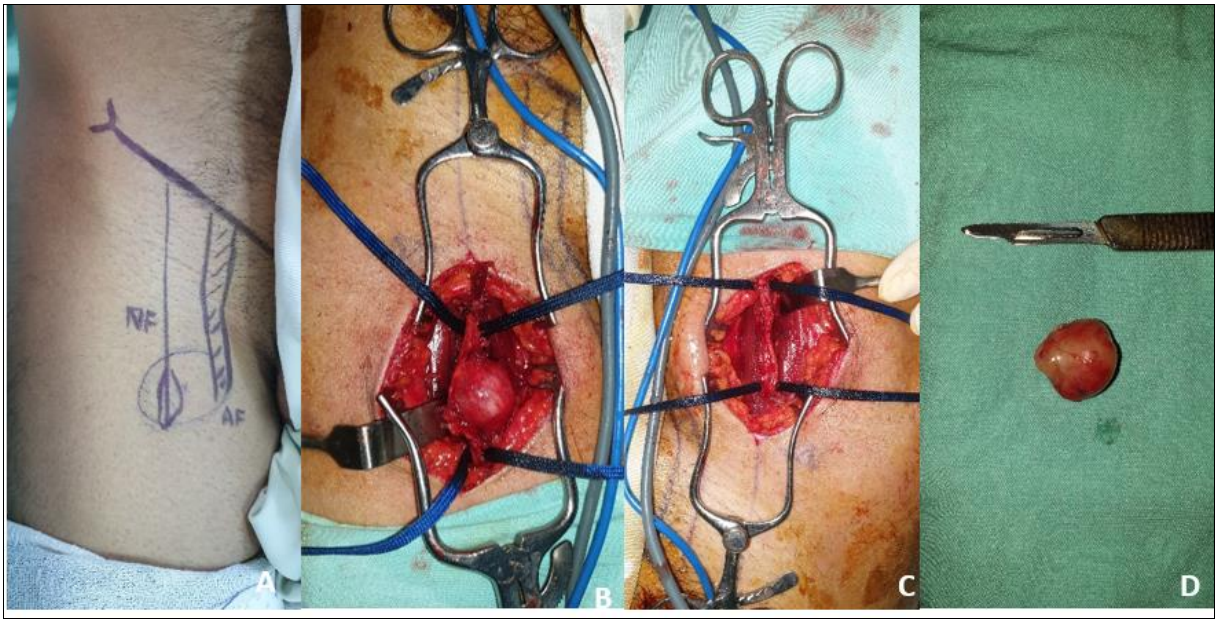

Fig 5: (A) Pre-operative mapping of the palpated tumour of the thigh. (B-C) Dissection a well-circumscribed firm tumour of the saphenous nerve. (D) Schwannoma after complete extirpation.

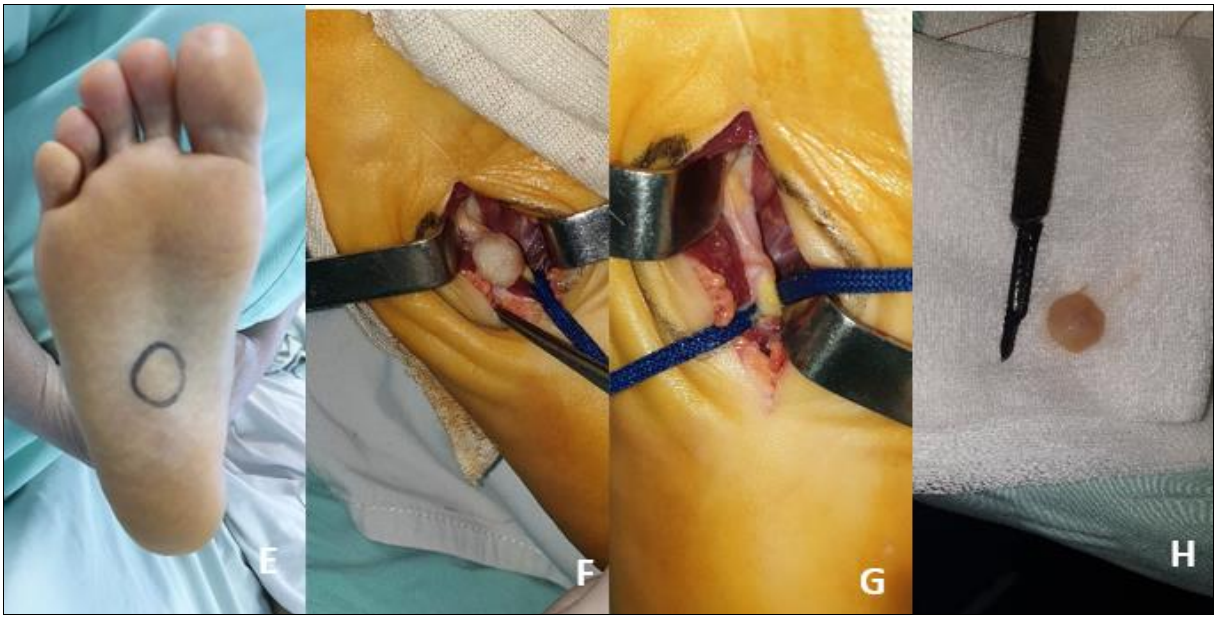

Fig 6: (E) Pre-operative localisation of the palpated mass. (F) Schwannoma of the medial plantar nerve. (F) Intact medial plantar nerve after complete tumour extirpation. (H) Schwannoma tissue after complete removal.

Discussion

Neurofibromatoses are a group of genetically-determined diseases characterised by a predisposition to develop tumours of the nerve sheath.

Neurofibromatosis type one (NF1), also known as von Recklinghausen's disease, accounts for 96% of all NF cases. It is an autosomal dominant hereditary disease, caused by mutations of the NF1 gene located on the 17q11.2 locus of chromosome 17. The most prevalent clinical manifestations of the disease are cutaneous "café-au-lait" macules, visible subcutaneous neurofibromas and "Lisch" nodules. Significantly associated to some malignant tumours and vascular disease, the life expectancy of NF1 patients is decreased by 15 years in comparison with the general population [1].

NF2, is a dominantly inherited disease. Affected individuals inevitably develop multiple schwannomas, the most common being bilateral vestibular nerve schwannomas, meningiomas and ependymomas. The majority of patients present with hearing loss or other symptoms in relation to vestibular nerve schwannomas, such as tinnitus or vertigo. [2] The disease is caused by a mutation of the gene that normally produces merlin, located at 22q12.2 of chromosome 22, and which regulated multiple proliferative signalling pathways [1].

Schwannomatosis is the term describing the clinicopathologic condition of multiple schwannomas lacking NF2 stigmata. It was first described by Shishiba *et al.* in 1973 [3]. Unlike solitary schwannomas, segmental schwannomatosis is uncommon. It is defined by the presence of multiple schwannomas of a deep major nerve in a single limb. Antinheimo *et al.* estimated the annual incidence of schwannomatosis to be 0.58 cases per 1,000,000 persons [4]. Seppälä reported that the prevalence of the disease was 3.3 % in a series conducted over the course of 10 years [5].

Diagnostic criteria for schwannomatosis were proposed by MacCollin *et al.* They required the association of two or more pathologically sampled schwannomas to the absence of a vestibular nerve tumor on radiology after the age of 18 to confirm the diagnosis. In the absence of cranial imagery, the presumptive diagnosis is based on the absence of symptoms of vestibular nerve dysfunction and the presence of either two or more pathologically confirmed schwannomas after the age of 30, or two or more confirmed schwannomas in an anatomically limited distribution at any age [6]. However recent studies, suggest that the presence of a unilateral vestibular schwannoma or intracranial meningiomas doesn't exclude the diagnosis of schwannomatosis [7].

Revised diagnostic criteria have been proposed by Baser *et al.* based on the United Kingdom registry of NF2 in order to increase the sensitivity and specificity of diagnosis and decrease the confusion and overlap between NF2 and schwannomatosis. In this study, definite schwannomatosis can be defined either by the presence of two or more non-intradermal schwannomas, at least one of which is histologically confirmed in a patient older than 30 years or by the presence of one pathologically confirmed schwannoma in a patient who has a relative who meets the latter criteria. The absence of signs of NF2, including vestibular schwannomas on high quality MRI scans or a known constitutional NF2 mutation and the absence of first-degree relatives with NF2 is essential for the diagnosis of definite and "possible schwannomatosis [8].

It has been recently established that inactivating germline mutations in the tumor suppressor genes SMARCB1 and LZTR1 are present in approximately 85% of families with Schwannomatosis and up to 40% of sporadic cases. Additional somatic mutations are required to cause the expression of the disease. The loss of heterozygosity of large parts of the chromosome 22q is responsible for the loss of the NF2 gene as well as the SMARCB1 and LZTR1 which triggers the disease [1].

Clinical presentation

Classically, the mean age at diagnosis was around 30 years with no particular sex predominance. However some authors have reported a female predominance in their series. [9, 10] Peripheral distribution of the tumors was characteristic in most series, with involvement of the lower limb in one third of the cases, the upper limb and the spine in 23% of the cases each, spine (23%), while other locations were noted in 19% of cases [11].

Our patient presented for the first time at the age of 29 with a schwannoma of the lower limb. The diagnosis of schwannomatosis was however confirmed later as the patient was operated on for the second and the third confirmed schwannomas of the same lower limb.

Chronic pain was the main clinical sign in more than 60% of the patients presenting with schwannomatosis, while the appearance of a mass was reported in 50 % of the cases [6]. Tingling, numbness and weakness were inconstantly present.

Unlike NF2 where the average age at death is 36 years, the life expectancy in schwannomatosis is normal [1].

Medical treatments such as pregabalin, opioids and anti-depressants were prescribed for pain control. Surgical resection was indicated for refractory pain and in cases of spinal cord compression or other organ impingement.

Conclusion

Schwannomatosis remains to this day unclearly understood owing in part to its low incidence. It can be functionally debilitating for young active adults. Surgical treatment remains the standard of care for accessible tumors, as no targeted therapy has yet to prove its efficacy.

Disclaimer: The patient has provided informed consent for publication of the case.

Conflict of Interest

Not available

Financial Support

Not available

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