

Von Recklinghausen disease (neurofibromatosis type 1): beyond skin involvement

Sara M Rocha, Maria Betânia Ferreira, Rosa Ribeiro, João Correia

Department of Internal Medicine, Centro Hospitalar Porto, Porto, Portugal

Correspondence to
Dr Sara M Rocha,
saraisabelrocha@gmail.com

DESCRIPTION

A 35-year-old black woman with Von Recklinghausen disease (or neurofibromatosis type 1) presented multiple *café-au-lait* spots, axillary freckling and multiple cutaneous neurofibromas, which were more expressive in the abdomen and trunk (figure 1). In the course of the disease, she developed nodular and diffuse plexiform neurofibromas, associated with mild pain and skin disfigurement (figure 1D). A CT scan was performed when she complained of lumbar pain, although she had a normal neurological examination. This study showed a heterogeneous lumbar mass involving the neurological structures and the electrophysiological examination proved a decreased conduction velocity in the left lower limb, supporting the diagnosis of neuropathy.

According to her disease, this proximal nerve hypertrophy could have two major aetiologies—a benign internal complex plexiform neurofibroma or a malignant peripheral nerve sheath tumour (MPNST). Other genetic diseases, such as Charcot-Marie-Tooth disease, Dejerine-Sottas disease and Noonan disease, were considered and excluded considering the absence of personal and family history suggestive of those diseases. Acquired causes of neuropathy, like chronic inflammatory demyelinating polyradiculoneuropathy, were also ruled out after several studies (haemoglobin, erythrocyte sedimentation rate, fasting morning glucose levels, serum urea and creatinine, electrolytes, liver function, serum immunoelectrophoresis, serum immunophenotyping, antinuclear antibody, thyroid-stimulating



Figure 1 Patient photography's: (A) skin-coloured soft papules and nodules, consistent of multiple cutaneous neurofibromas in the back, which increased in size and number with age; (B) flat uniformly hyperpigmented macules known as "café-au-lait" spots are also visible in the abdomen; (C) a subcutaneous neurofibroma on the right hand, presented as a firm and tender nodule; (D) a diffuse plexiform neurofibroma on the right lower member representing an important cause of morbidity and disfigurement.



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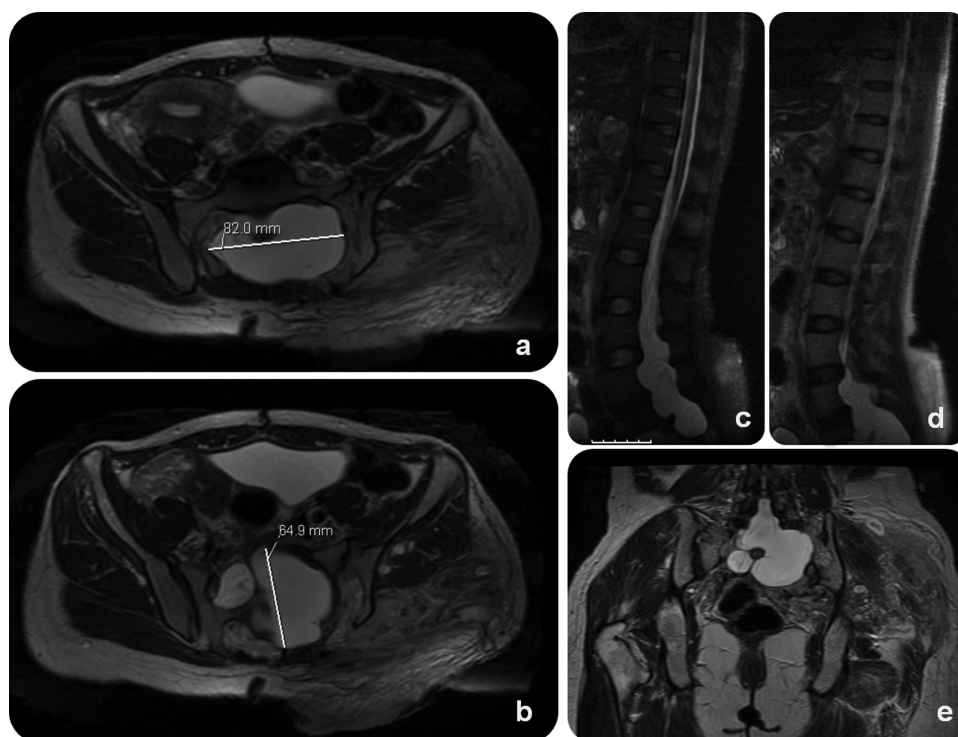


Figure 2 MRI in T2-weighted images: axial scans (A and B) showing a 65×82 mm hyperintense mass, suggestive of a multiplex neurofibroma; in sagittal (C and D) and coronal (E) scans, heterogeneous and infiltrative features were more evident, leading to the hypothesis of a malignant transformation.

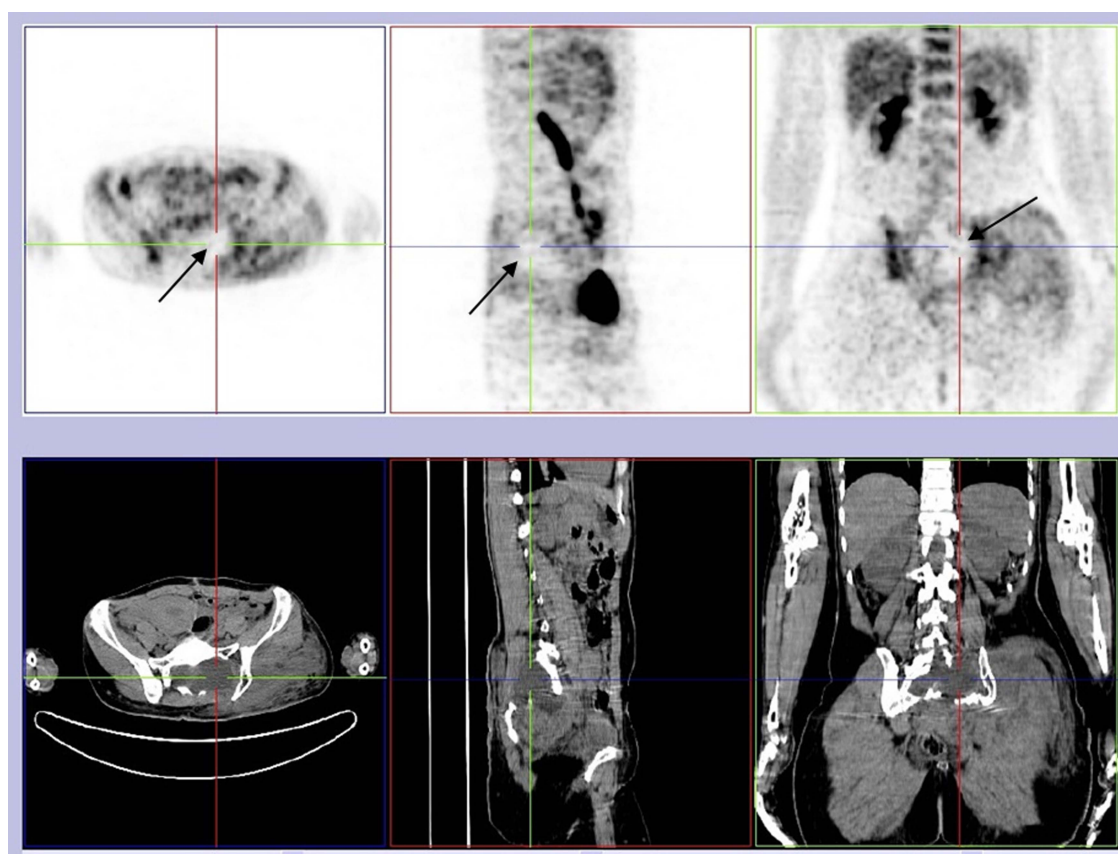


Figure 3 The F-18 fluorodeoxyglucose-positron emission tomography scan images showing a poor tracer uptake by the lumbar mass (signaled by the black arrow), suggesting a benign aetiology such as an internal complex plexiform neurofibroma.

hormone, vitamin B₁₂ and folate serum levels, serological studies for HIV, hepatitis B and C). The genetic testing for hereditary motor and sensory neuropathy type 1 was not performed.

MRI confirmed a huge and infiltrative mass (figure 2), poorly delimited and located on the left sciatic nerve route, extending from the sacroiliac region to the gluteus muscles (hyperintense lesion in T2); several small nodules could be seen inside, hypointense in T1 and hyperintense in T2 and were suggestive of neurofibromas. After contrast, the mass had heterogeneous uptake and infiltration of the subcutaneous adjacent tissue was seen. Although this lesion could be a plexiform neurofibroma with oedema and muscular denervation, it showed an atypical and aggressive behaviour and a malignant transformation into a MPNST could not be excluded. Considering the high surgical risk of the location, the biopsy was delayed and a positron emission tomographic scan was performed. In this examination, the mass had a poor uptake of F-18 fluorodeoxyglucose (figure 3), indicating a lower level of inflammation. Since this behaviour is not typical of proliferating aggressive tumours (which shows an enhanced tracer uptake), a malignant transformation was unlikely. Thus, a benign aetiology was considered in the form of an internal complex plexiform neurofibroma.

The patient was maintained under medical management, with pain control and radiological surveillance. Two years later this complex plexiform neurofibroma remains stable, without significant growth and without neuropathy progression, as described in the literature¹ but a continuous and close surveillance remains absolutely necessary.

Learning points

- ▶ Plexiform neurofibromas (PNs) are benign tumours arising from Schwann cells, but they can be locally aggressive (in 64%), causing pain and functional impairment.^{1 2}
- ▶ Internal PNs have more risk of malignant transformation into malignant peripheral nerve sheath tumour (signalled by a rapid increase in tumour size, worsening pain and new neurological deficits) and can be detected only by images studies.^{2 3}
- ▶ The management of these complex lesions is unclear; the surgical approach is particularly difficult because they are extensively infiltrative, highly vascularised and tend to recur.^{2 3}

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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