Neurofibromatosis type 1: focal dystonia associated with a malignant peripheral nerve sheath tumour—a video-illustrated case

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DESCRIPTION

A man aged 63 years was diagnosed with neurofibromatosis type 1 (also known as von Recklinghausen disease) at the age of 16. Over the years, he developed multiple plexiform neurofibromas throughout his skin, more expressive in the trunk (figure 1) as well as deep inside the body (figure 2). He was admitted to our ward with severe lumbar pain, weakness of both legs and faecal and urinary incontinence. On examination, he was found cachectic in appearance and paraparesis was

confirmed. He also presented focal dystonia of abdominal muscles characterised by involuntary muscle contractions (video 1).

Figure 1 Patient's photography: multiple nodular plexiform neurofibroma in the trunk.

Video 1 Patient's abdominal muscles involuntary contractions.

Figure 2 MRI images: superficial and deep plexiform neurofibroma (arrows) with the characteristic appearance of hyperintense mass.

Figure 3 CT scan showing a huge and heterogeneous lumbosacral mass (highlighted by arrows).

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A CT scan was performed and revealed a huge heterogeneous lumbosacral mass (figure 3). The lesion presented a rapid growth with compression and infiltration of surrounding struc-

tures and bone destruction, an atypical and aggressive behaviour for a plexiform neurofibroma. This suggested a malignant transformation into a malignant peripheral nerve sheath tumour. The patient presented a rapid and unfavourable evolution, passing away a month after admission.

Learning points

- ▶ Patients with NF1 and internal plexiform neurofibromas were 20 times more likely to have malignant peripheral nerve sheath tumours (MPNSTs), compared with individuals without internal plexiform neurofibromas.¹
- ▶ Pain and functional impairment are typical signs of MPNST, but dystonia due to a neurogenic tumour is extremely rare,² with few cases described.
- ▶ MPNSTs have a poor prognosis. With this in mind, individuals with NF1 and plexiform neurofibromas warrant increased surveillance for development of MPNST.³ Early detection should be sought in order to allow surgical treatment.

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Competing interests None declared.

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