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Case Report

A spinal cord compression syndrome revealing neurofibromatosis type 1: A case report ☆

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ABSTRACT

Neurofibromatosis type 1 (NF1), formerly known as von Recklinghausen disease is an autosomal dominant disease with multisystem involvement. In the peripheral nervous system, it leads to the development of benign tumors from the tissue of the spinal or cranial nerve sheaths, known as "neurofibromas." We report the case of a 40-year-old patient with spinal cord compression syndrome in whom spinal MRI revealed cervical, dorsal and lumbosacral neurofibromas revealing neurofibromatosis type 1.

Introduction

Neurofibromatosis type 1 (NF 1) is a genetic disease caused by a mutation in the neurofibrin gene located on chromosome 17q12. It is inherited in an autosomal dominant mode, although there are many cases of de novo mutation [1]. The clinical manifestations are polymorphic and affect different organs. However, involvement of the peripheral nervous system, from which neurofibromas and plexiform neurofibromas develop, remains the most frequent and specific manifestation of the disease. We report the case of a 40-year-old man with neurofibromatosis type 1 secondary to a de novo mutation, revealed by a spinal cord compression pattern.

Case report

A 40-year-old man, with no personal or family history of genetic disease, consulted for progressively worsening low back pain for more than 2 years. Physical examination revealed multiple nodular lesions on the trunk and along the upper limbs, with "café au lait" spots larger than 15 mm on the thorax, associated with ephelides (Fig. 1). Neurological assessment revealed a spinal cord compression syndrome consisting of tetra pyramidal syndrome, paresthesias and neurological claudication. There were no visual disturbances or intracranial hypertension syndrome. Brain and spinal cord MRI were performed. The cerebral level showed intraorbital, extra conical

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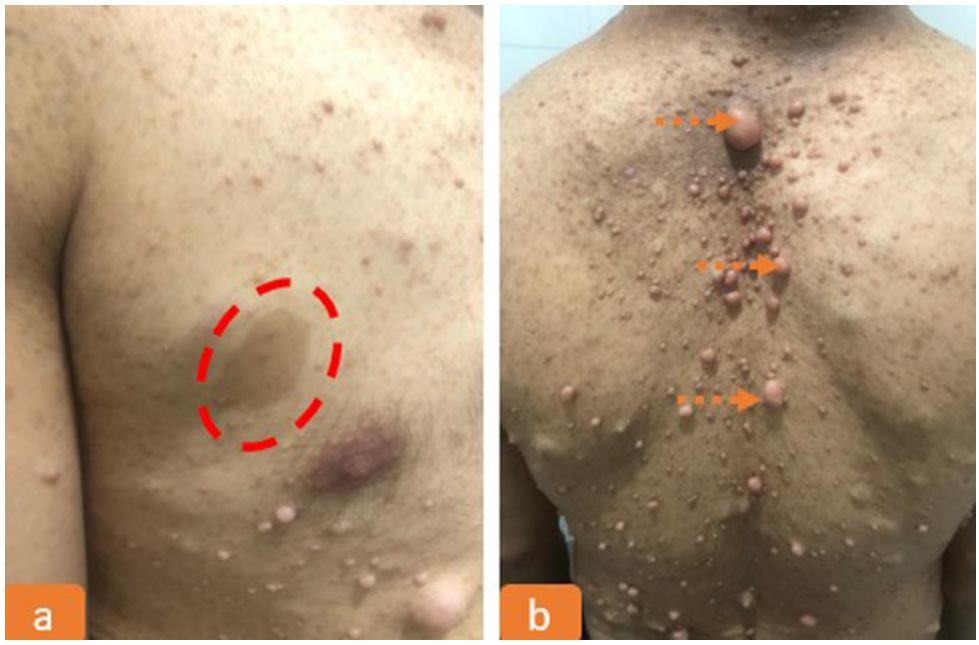

Fig. 1 – Ventral (A) and dorsal view of the thorax showing a large « café au lait macule » ( ) and cutaneous neurofibromas ( ).

expansive processes of bilateral topography with an intermediate signal on T1 and T2 weighted images enhanced after injection of contrast product, suggestive of bilateral neurofibromas of the branches of the V1 nerve (ophthalmic branch of the trigeminal nerve). There were no abnormalities of the optic nerves, brain parenchyma or meninges. In the spine, there was scoliosis of the cervical spine with multiple expansive processes centered on the cervical, dorsal and lumbosacral nerve roots, responsible for a staged enlargement of the intervertebral foramens related to neurofibromas (Fig. 2). In the cervical region, these neurofibromas exerted a mass effect on the closed bulb and cervical cord, which were pushed backwards, explaining the patient's symptoms (Fig. 3). The patient's management was the subject of a multidisciplinary concertation and the decision to perform a decompressive laminectomy was adopted.

Discussion

Neurofibromatosis type 1 or von Recklinghausen disease is the most common phacomatosis with an incidence of approximately 1 case per 3000 newborns [2]. It is an autosomal dominant disease that affects both the peripheral and central nervous system as well as other organs such as the skin and musculoskeletal tissue, which can lead to alteration of the quality of life of patients. The diagnostic criteria are nowadays well defined (Table 1) [3], so that the disease is mostly detected in childhood. However, in some cases, it is diagnosed in adulthood when the lesions worsen. Multiple clinical manifestations are found in neurofibromatosis type 1. The main organs or systems affected are [4]:

- Skin: « Café au lait » macules (brown skin spots), ephelides, subcutaneous neurofibromas, iris hamartomas or lisch nodule
- Bones: peripheral bone deformity with limb deformities, spinal involvement (spinal scoliosis, vertebral scalloping, enlargement of intervertebral foramina)
- The nervous system.

In the peripheral nervous system, neurofibromas and plexiform neurofibromas are the most frequently observed in neurofibromatosis type 1. They can occur at any level and are seen on imaging as well-limited (for neurofibromas) and poorly limited (for plexiform neurofibromas) masses with an intermediate signal on T1- and T2-weighted images, which enhance homogeneously after injection of contrast agent. These masses may be responsible for scalopping on the spine, dural ectasia, widening of the intervertebral foramina or even a slow spinal cord compression syndrome for the most voluminous ones as illustrated in our case. Plexiform neurofibromas can degenerate into neurofibrosarcomas with a poor prognosis. In the central nervous system, multiple gliomas (preferably gliomas of the optic tract), subependymal giant cell astrocytomas, or unidentified white matter bright objects may develop. Imaging plays an important role in the management of patients with NF1. In addition to contributing to the positive diagnosis, it allows an exhaustive lesion assessment and is used to detect and follow-up lesions of the central nervous system, but also to look for signs of degeneration of plexiform neurofibroma into neurofibrosarcoma, which must be suspected in the case

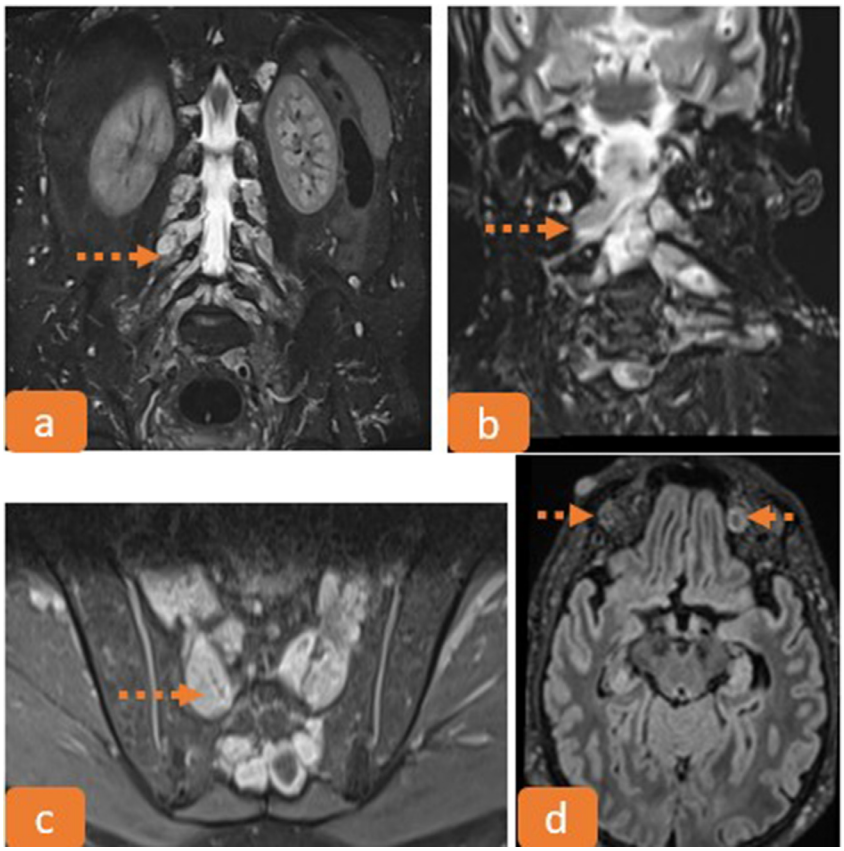

Fig. 2 – Spinal cord MRI in coronal STIR at the dorsolumbar level (A); in coronal STIR at cervical level (B); and axial T1 Fat suppression postcontrast enhancement through the sacroiliac joint (C) showing stepped neurofibromas (▶), enlarging the intervertebral foramina. Axial FLAIR (D): bilateral neurofibromas of the V1 branches are noted.

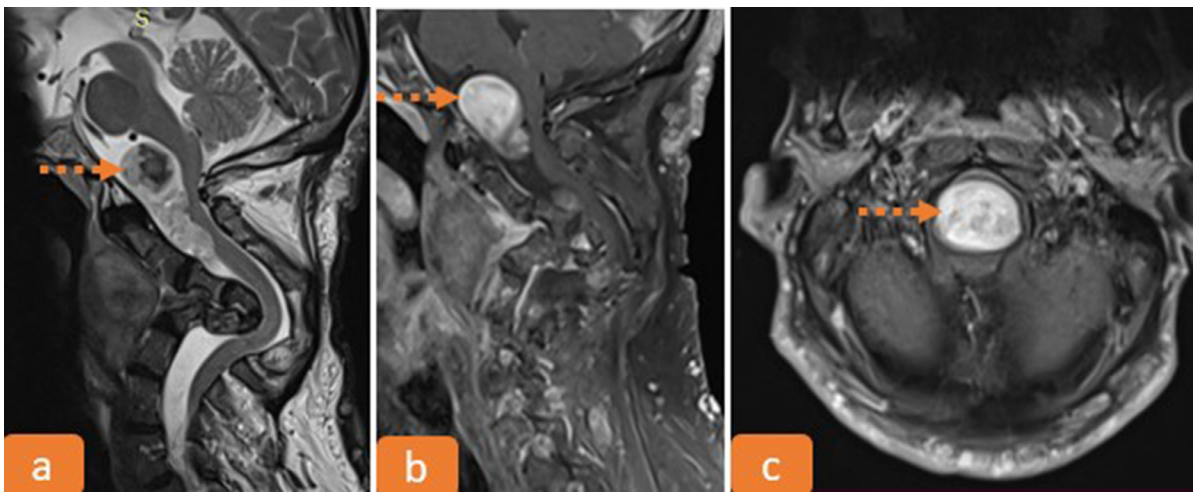

Fig. 3 – Cervical spinal cord MRI, sagittal T2-weighted image (A); T1 Fat suppression postcontrast enhancement in sagittal section (B) and axial section (C), showing a large neurofibroma compressing the closed bulb and the cervical cord (▶). A scoliotic deformation of the cervical spine is also noted.

Table 1 – Updated diagnostic criteria for neurofibromatosis type 1 [3].

√ At least 2 of the following criteria are necessary for the diagnosis
√ At least « six café » au lait spots ≥ 15 mm in adults or 5 mm in children*
√ Axillar or inguinal ephelides*
√ At least 2 neurofibromas or 1 plexiform neurofibroma
√ At least 2 Lisch nodules of the iris or 2 or more choroidal abnormalities
√ A glioma of the optic pathway
√ A specific bone lesion (tibial dysplasia; pseudarthrosis of a long bone; sphenoid dysplasia)
√ A pathogenic NF1 gene variant
√ First degree family history

* At least 1 of the 2 pigmentary findings (café-au-lait macules or ephilides) should be bilateral

of worsening of the symptomatology. There are 2 other types of neurofibromatosis: neurofibromatosis type 2, which is less common than type 1, and schwanomatosis, which is the rarest form. Neurofibromatosis type 2 is also a genetic disease (NF2 gene on chromosome 22) of dominant inheritance, with many cases of de novo mutation. It is responsible for the development of schwanomas of the cranial nerves, in particular vestibular swchanomas, but also other tumours: schwanomas of the spinal nerves, intracranial meningiomas and meningiomas of the spinal cord, as well as glial tumors (mainly ependymomas) [5]. Schwanomatosis, on the other hand, leads to overlapping manifestations with those of neurofibromatosis types 1 and 2 (meningiomas, schwanomas). However, it is a distinct disease that is frequently associated with mutations in the SMARCB1 or LZTR1 genes [6].

Conclusion

Neurofibromatosis type 1 is an autosomal dominant disease, although de novo mutations are not uncommon. This disease is responsible for polymorphic clinical manifestations and affects many organs, in particular the peripheral nervous system, from which neurofibromas develop and can increase in size and lead to compression of the spinal cord.

Patient consent

The patient's informed consent was obtained

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