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Case Report

Neurofibromatosis Type 1 Complicated by Slow Spinal Cord Compression: A Case Report

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ABSTRACT

Neurofibromatosis type 1 (NF1) is a genetic disease with multisystem involvement, mainly affecting neural crest-derived cells. It is a rare disease, with 5 cases per year. In Madagascar, its estimated prevalence is 0.39%. The most common complications reported in a Malagasy research series are learning disability, epilepsy and scoliosis. Slow NF1-related spinal cord compression is possible but rare, and few cases are reported worldwide. We report a slow spinal cord compression staged by cutaneous neurofibromas in a patient with NF1.

Keywords: Antananarivo; Cutaneous neurofibromas; Neurofibromatosis type 1; Spinal cord compression

Introduction

Von Recklinghaussen disease or neurofibromatosis type 1 is a genodermatosis described in 1882 by Friedrich Von Recklinghaussen. It is the most common neurofibromatosis. The disease mainly affects females at an early age. Its incidence is estimated at around 1/2000, and its prevalence at 1/2000 to 1/3000 according to European, North American and Oceanian studies[1,2,3]. In Madagascar, its prevalence was 0.39% in 2019[4]. NF1 is characterized by the presence of café au lait stains, lentigines and neurofibromas (cutaneous or plexiform). Other manifestations may be associated, such as Lisch nodules, spinal scoliosis, gliomas, neuroendocrine tumors and even, in rare cases, spinal cord complications (compression)[5]. Despite current therapeutic advances, the disease is still complicated by learning difficulties, epilepsy, scoliosis and spinal cord compression, altering patients quality of life[4,5]. We aim to report a case of NF1 complicated by slow spinal cord compression.

Case Presentation

Our patient was a 31-year-old man of Comorian origin hospitalized for a presentation of tetraparesis. His history noted a learning delay with a secondary school education, and he is currently out of work. There was no family history of neurofibromatosis.

His history was marked by the progressive development, over the past 1 year, of tetraparesis responsible for gait disorders. The recent onset of sphincteral disorders such as constipation and dysuria prompted hospitalization. No spinal trauma was noted.

Clinical examination revealed a conscious, apyretic patient in good general health. Neurological examination revealed paraparesis with muscle strength at 2/5, no sensory deficit, osteotendinous reflexes and muscle tone were abolished. Bladder and bowel examination revealed a bladder globe with overflow micturition and constipation requiring manual exoneration. Mucocutaneous examination revealed multiple cutaneous neurofibromas on the trunk and back, café au lait spots on the trunk and axillary lengitines (**Figure 1**).

MRI of the entire spine revealed multiple extramedullary lesions at cervical level, with T2 hypersignal and T1 hyposignal, and STIR hypersignal opposite C2 C3 C4 C5 C6 C7, compressing the medulla (**Figure 2**). In the lumbar region, disc dehydration

with heterogeneous hyposignal appearance in S2 S3 with disc pinching, multiple T2 hypersignal, T1 hyposignal and STIR hypersignal lesions extra-medullary along diffuse lumbosacral radicular paths compressing the cauda equina (**Figure 3**).

Biological examinations revealed no inflammatory syndrome. Phosphocalcium levels, serum protein electrophoresis and renal function tests were normal.

The diagnosis of NF1 complicated by spinal cord compression was made in view of the patient's presentation according to the diagnostic criteria of the French National Authority for Health (HAS), revised in 2021.

Surgical removal of the cervical neurofibromas was proposed but refused by the patient. Sphincter disorders were treated with an indwelling bladder catheter, laxatives and repeated manual exoneration. Motor rehabilitation was prescribed for his paraparesis.

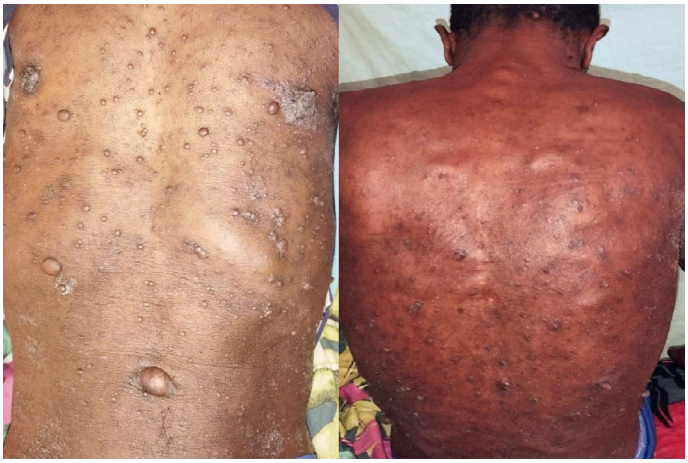

Figure 1: Multiple cutaneous fibromas with café au lait staining.

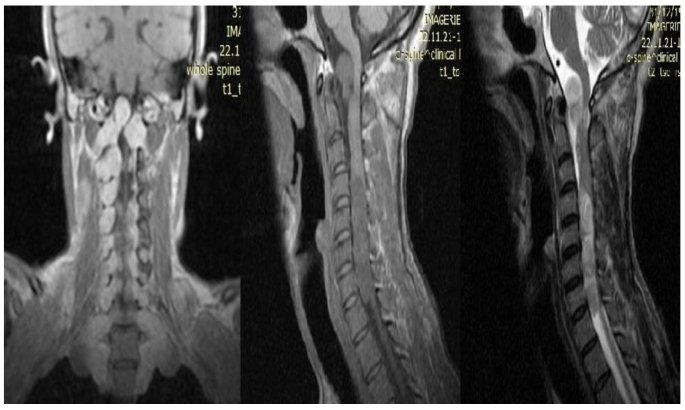

Figure 2: Extra medullary Compression by a Cervical Neurofibromas.

Discussion

Many cases of NF1 have been reported around the world, with the complication that patients' quality of life is impaired by the psychological effects of the sometimes spectacular lesions, and that children experience learning difficulties, as well as static and neurological disorders. The seriousness of the disease lies in the tumors, which can degenerate into malignant tumors. Apart from the cutaneous manifestations of NF1 (café-au-lait spots, lentigines), neurofibromas are unique[5,6].

Neurological manifestations due to tumours in NF1 are varied, and include central nervous system and peripheral nervous system disorders. Malignant tumours of the nerve sheaths are very rare before the age of 10, and constitute the main complication of NF1 in adulthood (3-5%)[5,8]. They can develop from plexiform, cutaneous or visceral neurofibromas. Neurofibromas are benign tumors that develop in a nerve or nerve sheath, causing sensory disturbances and motor deficits[7,9]. They develop late in childhood or very early in adolescence. Because they are rich in mast cells, neurofibromas are pruritic, which explains the discomfort associated with the appearance of the lesion. Sensory-motor deficits are associated with damage to spinal nerve sheaths[9,12,13].

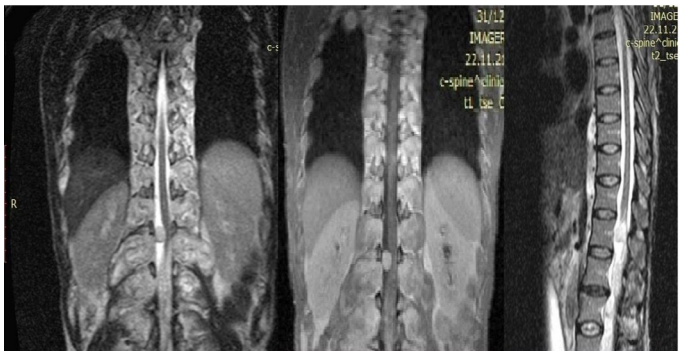

Figure 3: Extra medullary Compression by a Cervical Neurofibromas in the Lumbar Spine.

Plexiform neurofibromas are present in 30-50% of individuals with NF1. They usually develop at birth, then in adolescents and young adults. They are very large in appearance, frequently involving nerve sheaths and invading underlying structures. These plexiform neurofibromas are at high risk of malignant degeneration[10,11].

Other structural damage is also present in NF1, including osteopenia, scoliosis, sphenoidal dysplasia, pseudarthrosis and congenital tibial dysplasia. Patients with NF1 are generally short for their age and have low bone mineral density. The risk of fracture is higher in adults with NF1 than in kids with NF1[10,11,12,16].

For peripheral or central neurological involvement, magnetic resonance imaging is the most sensitive for visualizing peripheral spinal cord involvement associated with neurofibromas, whether malignant or benign. Computed tomography (CT) is useful for looking for tumour invasion or for tumours in the central nervous system[13,14].

As far as treatment of neurological damage is concerned, surgical excision is always indicated to limit and control the symptoms of compression. Secondarily, depending on the malignant or benign nature of neurofibromas, chemotherapy is required for those that are malignant[6,14].

Several therapies are currently being tested to improve the prognosis and quality of life of NF1 patients[6].

Conclusion

NF1 is a disease that can be complicated by severe and disabling sensory-motor disorders. These complications are linked to damage to the nerve sheaths of cutaneous or plexiform neurofibromas. The most serious is spinal cord compression. Decompressive surgery is always indicated, in conjunction with chemotherapy if malignant.

Conflicts of Interest

We do not have any conflict of interest.

Consent Statement

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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