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Case Report

Familial Spinal Manifestations of Neurofibromatosis Type 1 : A Report of Two Cases

Manifestations Spinales Familiales d'une Neurofibromatose Type 1 : À Propos de Deux cas

Julie Marie Adeline W Kyelem[1.5], Yakouba Haro[2.5], Madi Kam[3], Siguiya Abraham W Zaongo[4], Alfred Anselme Dabilgou[1.5], Djingri Labodi D Lompo[5], Christian Napon[5], Athanase Millogo[5]

Affiliations
1. Neurology Department, Yalgado Ouedraogo University Hospital, Ouagadougou
2. Neurosurgery Department, Yalgado Ouedraogo University Hospital, Ouagadougou
3. Pediatrics Department, Charles de Gaulle University Hospital, Ouagadougou
4. Radiology Department, Yalgado Ouedraogo University Hospital, Ouagadougou
5. Neurology Department of the Unité de Formation et de Recherche en Sciences de la Santé, Joseph KI-ZERBO University, Ouagadougou.

Corresponding Author
Kyelem Julie Marie .Adeline .Wendlamita
Tel: (00226)62 83 33 62
Email: linejulyk@gmail.com

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ABSTRACT

Neurofibromatosis type 1 (NF1) is a common inherited autosomal dominant disease. The most frequent manifestations are cutaneous and neurological. We report a case of familial deforming neurofibromatosis type 1 in a mother and daughter causing slow cord compression. The mother had consulted the neurology department of the Centre Hospitalier Universitaire Yalgado Ouédraogo (CHU-YO) for the first time in 2017, with progressively worsening weakness of all four limbs in a context of progressive spinal deformity. For 14-year-old schoolgirl, daughter of patient 1, she had been suffering from skin lesions for 5 years. She had been seen for recently worsening back pain (10 months ago), marked by the onset of spinal deformity and difficulty walking, with a notion of intermittent spinal claudication. This condition was associated with diffuse skin lesions that had previously prompted a dermatology consultation, and the symptomatology had progressed to motor deficit in all four limbs. Spinal cord magnetic resonance imaging was used to diagnose the nerve lesions. The severity of spinal lesions requires clinical monitoring for early detection of neurological symptoms, especially disabling spinal manifestations. Our case highlights the importance of establishing clinical surveillance in NF1 patients. MRI is the examination of choice for early detection of nerve lesions.

RÉSUMÉ

La neurofibromatose de type 1 (NF1) est une maladie héréditaire autosomique dominante fréquente. Les manifestations les plus fréquentes sont cutanées et neurologiques. Nous rapportons un cas de neurofibromatose déformante familiale de type 1 chez une mère et sa fille provoquant une compression médullaire lente. La mère avait consulté le service de neurologie du Centre Hospitalier Universitaire Yalgado Ouédraogo (CHU-YO) pour la première fois en 2017, pour une faiblesse des quatre membres d'aggravation progressive dans un contexte de déformation rachidienne progressive. Pour la collégienne de 14 ans, fille de la patiente 1, elle souffrait de lésions cutanées depuis 5 ans. Elle avait consulté pour des douleurs dorsales d'aggravation récente (10 mois auparavant), marquées par l'apparition d'une déformation de la colonne vertébrale et d'une difficulté à marcher, avec une notion de claudication intermittente de la colonne vertébrale. Cette pathologie était associée à des lésions cutanées diffuses qui avaient déjà motivé une consultation en dermatologie, et la symptomatologie avait évolué vers un déficit moteur des quatre membres. L'imagerie par résonance magnétique de la moelle épinière a permis de diagnostiquer les lésions nerveuses. La gravité des lésions spinales nécessite un suivi clinique pour la détection précoce des symptômes neurologiques, en particulier des manifestations spinales invalidantes. Notre cas souligne l'importance d'établir une surveillance clinique chez les patients atteints de NF1. L'IRM est l'examen de choix pour la détection précoce des lésions nerveuses.

INTRODUCTION

Neurofibromatoses (NF) are autosomal dominant genetic disorders. Neurofibromatosis type 1 (NF1) is the most common NF, the criteria for which were established at the 1987 National Institute of Health consensus conference [1, 2]. The first diagnostic criteria were established in 1988. The diagnosis of NF1 is made in an individual with 2 of the following clinical features:(1) hyperpigmented macules; (2) intertriginous freckles; (3) Lisch nodules;(4) neurofibromas; (5) optic tract gliomas; (6) optic tract gliomas (OPG); (7) distinctive bone lesions; and (8) a first-degree relative with NF1 [3, 4].

The existence of spinal lesions is associated with a major criteria suggestive of NF1 in the revised version of the 2021 consensus [5, 6, 7]. We report the cases of a mother and her daughter presenting with a severe form of NF1 with spinal locations.

OBSERVATION

Clinical case 1

This was a 37-year-old, single mother residing in Ouagadougou. She consulted the neurology department of the Centre Hospitalier Universitaire Yalgado Ouédraogo (CHU-YO) for the first time in 2017, with progressively worsening weakness of all four limbs in a context of progressive spinal deformity. This condition was associated with diffuse skin lesions that had previously prompted a dermatology consultation, and the symptomatology had progressed to motor deficit in all four limbs. The patient's history was unremarkable.

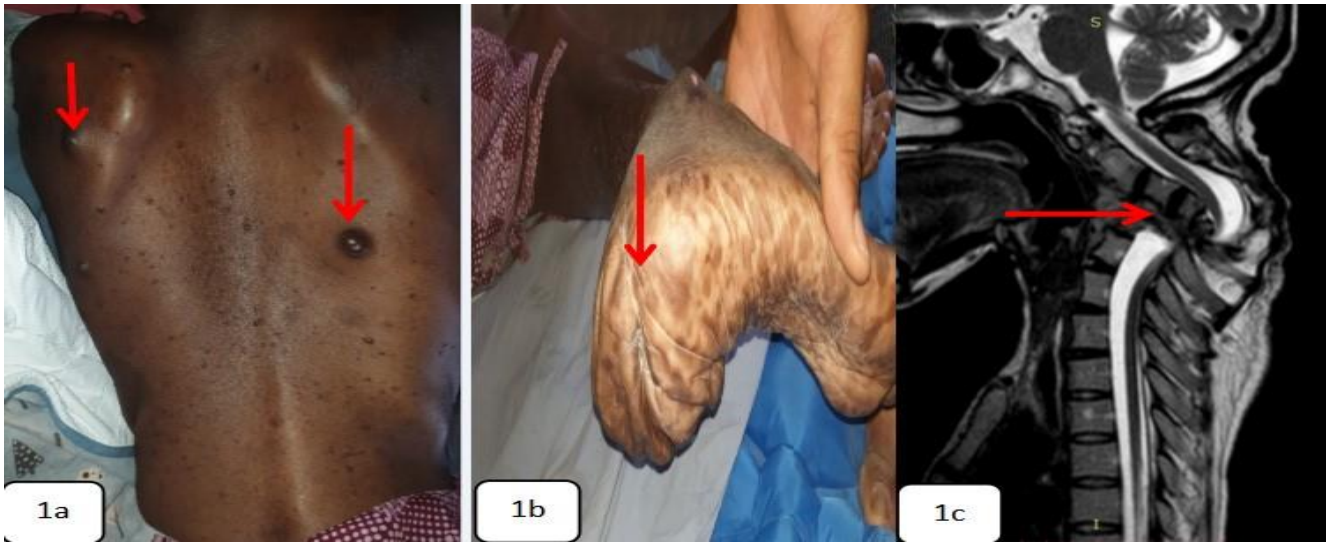

Figure 1. Photograph showing hyperpigmented macules «café-au-lait» macules, diffuse neurofibromas(1a). Plantar lentigines (1b). Cervical MRI sagittal section T2 showed spinal cord compression(1c).

Clinical examination revealed

Poor general condition, WHO stage 3. Spastic tetraparesis rated 3/5 on the Medical Research Council (MRC) scale, sharp osteotendinous reflexes and pyramidal hypertonia in all four limbs. Superficial tact hypoesthesia ascended to D4 dermatome. Sphincter hypertonia was associated with urine retention and constipation.The skin showed diffuse hyperpigmented macules, diffuse neurofibromas (figure 1a), palmoplantar lentigines (figure 1b). Examination of the spine revealed cervical gibbosity and cervical cyphosis.

Paraclinical examinations

MRI of the spinal cord revealed: sagittal section: significant deformation of the cervical spine with gibbosity, leading to severe compression of the spinal cord (as evidenced by loss of continuity of the T2 hypersignal of the CSF); sagittal sections, T2 sequence, STIR and T1+ Gado, revealed abnormal hypersignal, in STIR with enhancement after injection, attesting to the associated inflammatory phenomenon (figure 1c).

Clinical case 2

This case involved a 14-year-old schoolgirl, daughter of patient 1, who had been suffering from skin lesions for 5 years. She had been seen for recently worsening back pain (10 months ago), marked by the onset of spinal deformity and difficulty walking, with a notion of intermittent spinal claudication. This symptom evolved in an apyretic context and without convulsion.

Clinical examination revealed

General condition preserved, with limited mobility.
Neurological examination: motricity examination revealed paraparesis of the lower limbs rated 4/5 (MRC) proximally and right brachial monoparesis with sharp osteotendinous reflexes in the lower limbs, and bilateral Babinski's sign. Sensory examination revealed poorly systematized superficial hypoesthesia and tuning fork hypoesthesia, predominantly in the lower limbs, in all modes.examination of the meninges, cranial nerves and upper functions was normal.there was an absence of convulsive seizures. Examination of the skin and skin appendages revealed hyperpigmented macules on the right wrist, right elbow, left elbow and palmar lentigines, as well as cutaneous neurofibromas on the dorsal surface of the right shoulder (figure 2a). Examination of the skin and appendages revealed hyperpigmented macules of varying size on the right wrist, right elbow on the anterior surface, left elbow and palms of the hands, and cutaneous neurofibromas on the dorsal surface of the right shoulder (Figure 2a).

Paraclinical examinations

A CT scan of the spine revealed a dextroconcave scoliosis-like deformity of the dorsal spine in a 14-year-old girl, with a COBB angle greater than 30° (Figure 2b).

Spinal cord MRI T1 showed plexiform neurofibromas involving nerve roots (Figure 2c), and cerebral angiography was normal. The diagnosis of neurofibromatosis type 1 was made in both mother and daughter, with the presence of more than 4 clinical criteria in each case.

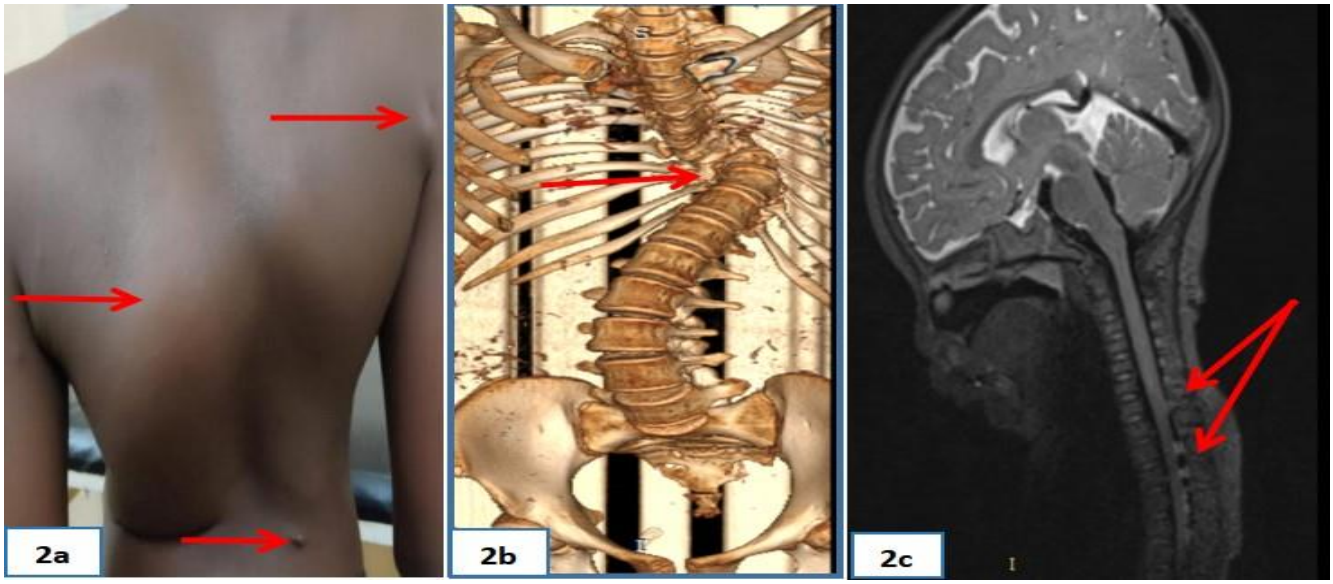

Figure 2. (2a) Photograph showing dorsal scoliosis, a left dorsal gibbosity and a cutaneous neurofibroma located on the dorsal right shoulder. (2b):CT scan of the spine in sagittal window section with 3D reconstruction showing scoliotic deformity of the dorsolumbar spine. (2c): MRI: sagittal T1 section showing nerve root involvement with plexiform neurofibromas in the 14-year-old patient.

DISCUSSION

Neurological lesions in NF1 are multifaceted and include, among others, central nervous system lesions, which contribute to the severity of the disease[8]. Sendrasoa et al found spinal involvement in 39.30% of cases[9]. Many bone lesions have also been described, including progressive thoracic scoliosis and vertebral dysplasia[10]. The peripheral nervous system is also the site of development of superficial cutaneous or peripheral nodular neurofibromas, or plexiform neurofibromas in nerve trunks and sheaths. Plexiform neurofibromas may be the source of clinical radicular symptomatology; they are ovoid, homogeneous tumours that develop long nerve tracts, and may be accompanied by intracanal extension [10, 11]. Scoliosis is a frequent spinal deformity in NF1, apart from nerve damage [11, 12, 13]. Spinal nerve damage impairs patients' quality of life, and neurosurgical management is difficult due to the incidence of surgical complications such as infection, haematoma, cerebrospinal fluid leakage and meningitis [14, 15]. Our case highlights the importance of establishing clinical surveillance in NF1 patients. MRI is the examination of choice for early detection of nerve lesions.

CONCLUSION

NF1 is a frequent hereditary disease. The wide clinical variability and frequency of neurological involvement necessitate clinical monitoring and appropriate paraclinical examinations. Spinal damage is a frequent cause of deformity and disabling spinal cord injury.

Conflict of Interest

We have no conflicts of interest.

Data Availability

All data from this study are available from the corresponding author under a reasonable request.

Consent

Maternal consent has been obtained

Authors' Contributions

Julie Marie Adeline W Kyelem, Yakouba Haro, Madi Kam and Siguiya Abraham W Zaongo contributed to the data analysis and writing of the article. Alfred Anselme Dabilgou, Djingri Labodi D Lompo, Christian Napon and Athanase Millogo supervised the writing.

Ethical Considerations

Our study is not unethical

We have no source of funding

We confirm that the current manuscript is not submitted to other journals.

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