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Retroperitoneal Manifestations of Type 1 Neurofibromatosis: A Case Report

A. Elmoctar[1]*, M. Ouazine[1], H. Bello[1], K. Rebbouh[1], Y. Sakhy[1], H. Tabakh[1], A. Siwane[1], N. Touil[1], O. Kacimi[1]

[1]Emergency Radiology Department, Ibn Rochd University Hospital, Casablanca, Morocco

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***Corresponding author:** A. Elmoctar
Emergency Radiology Department, Ibn Rochd University Hospital, Casablanca, Morocco

Abstract | **Case Report**

Type 1 neurofibromatosis (NF1) is a common autosomal dominant genetic disorder characterized by various manifestations, including plexiform neurofibromas; retroperitoneal involvement is rare but can complicate diagnosis and management. We report the case of a 42-year-old man presenting with pelvic pain and a left gluteal mass. Clinical examination revealed café-au-lait macules, cutaneous nodules, and Lisch nodules, and imaging showed nodular formations surrounding the bladder and rectum; a biopsy confirmed the presence of benign plexiform neurofibromas. Retroperitoneal neurofibromas in NF1 have an incidence of 1 to 3% and are often asymptomatic, although their growth may lead to local compression and clinical symptoms; identification through imaging and histological confirmation are essential for guiding management, which is often limited to monitoring and symptomatic treatment. NF1 is a disease with diverse manifestations requiring regular follow-up, and although rare, retroperitoneal neurofibromas play a diagnostic role and necessitate appropriate management.

Keywords: Type 1 Neurofibromatosis, Plexiform Neurofibroma, Retroperitoneal Manifestations, Von Recklinghausen Disease, Lisch Nodules, Case Report.

INTRODUCTION

Type 1 neurofibromatosis (NF1), or Von Recklinghausen's disease, is a common hereditary disorder with autosomal dominant inheritance, characterized by significant clinical variability, including neoplastic and non-neoplastic manifestations (Jett & Friedman, 2010). Approximately 50% of cases result from a sporadic mutation due to the high mutagenicity of the NF1 locus (Basile *et al.,* 2010). Among its rare but characteristic forms, plexiform neurofibromas develop from spinal nerves and can affect various anatomical regions such as the skin, paravertebral structures, and retroperitoneum (Tchernev *et al.,* 2016). Here, we present the case of an NF1 patient with a retroperitoneal plexiform neurofibroma, illustrated by pelvic imaging and associated manifestations.

CASE PRESENTATION

Mr. B.S, a 42-year-old man, presented to the emergency department with proctalgia and a sensation of pelvic heaviness. Clinical examination revealed a protruding, soft mass deforming the left buttock (Fig. 1A). The patient also had multiple café-au-lait macules of varying sizes and firm cutaneous nodules located on the back, axillae, and thoracoabdominal regions (Fig. 1B).

Ophthalmological examination revealed pigmented hamartomas of the iris consistent with Lisch nodules. No surgical history was reported, and routine biological tests and endoscopy were normal.

CT scan (Figure 2) and MRI (Figure 3) showed:

- Retroperitoneal nodular formations in the mesorectal and mesosigmoid fat. These formations were confluent, forming a pseudo-pelvic mass surrounding the bladder, rectum, and part of the sigmoid colon.
- Diffuse cutaneous neurofibromas.
- Lumbar scoliosis associated with dural ectasia at L4–L5.

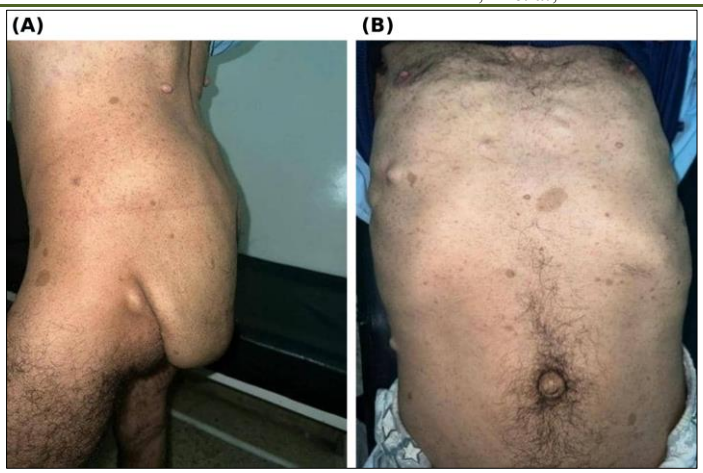

Figure 1: (A) Protruding, soft mass deforming the left buttock. (B) Multiple café-au-lait spots and cutaneous nodules present on the back and abdomen

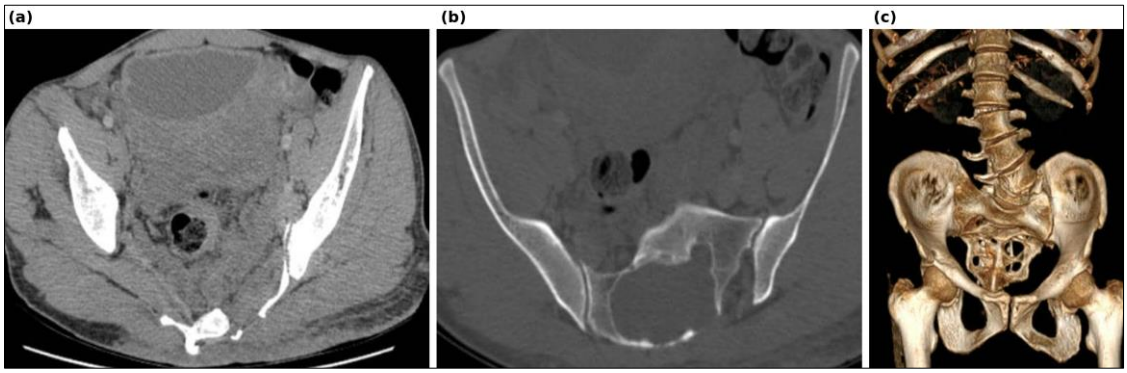

Figure 2: Abdominal CT scan in axial sections in parenchymal window (a) and bone window (b) and 3D reconstruction (c); nodular retroperitoneal masses isodense with minimal enhancement. No cavitation or calcification. Note the scoliosis on 3D reconstruction (c)

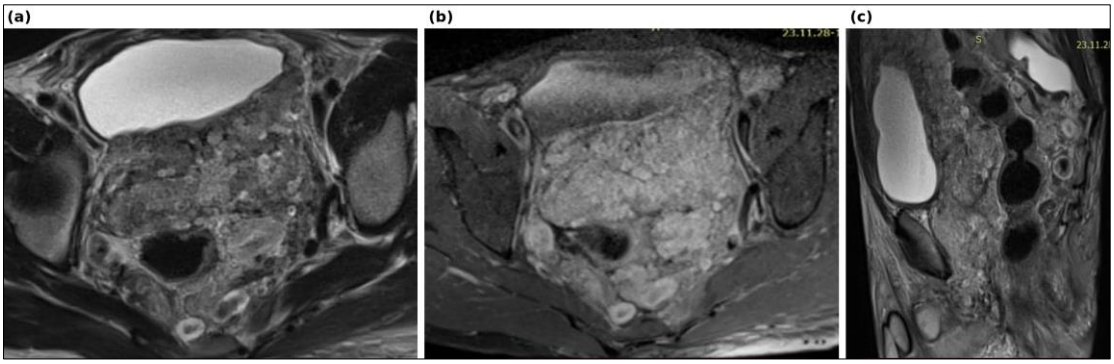

Figure 3: Pelvic MRI in axial T2 (a), T1 after gadolinium injection (b), and sagittal T2 (c): Heterogeneous retroperitoneal mass, predominantly hyperintense in T2 sequences with a hypointense center ("ring" appearance) with slight central enhancement in T1+ gadolinium sequence

A biopsy of the subperitoneal nodules and subcutaneous mass confirmed the diagnosis of benign plexiform neurofibromas, deemed inoperable. Management was limited to regular clinical follow-up and symptomatic treatment with analgesics.

DISCUSSION

NF1 is an autosomal dominant genetic disorder caused by a mutation in the NF1 gene located on chromosome 17q11.2. It is one of the most common genetic diseases, with an incidence of 1/2500–3000 (Victorio, 2020).

Diagnosis is based on at least two of the diagnostic criteria established during the 1988 consensus conference (Jett & Friedman, 2010):

- Six or more café-au-lait macules over 5 mm in diameter in prepubertal individuals and over 15 mm in postpubertal individuals.
- Two or more neurofibromas of any type or one plexiform neurofibroma.

- Freckling in the axillary or inguinal regions.
- Optic glioma.
- Two or more Lisch nodules (iris hamartomas).
- A distinctive osseous lesion such as sphenoid dysplasia or thinning of the long bone cortex with or without pseudoarthrosis.
- A first-degree relative with NF1.

The first symptoms often appear within the first year of life, with café-au-lait spots. Other signs, such as freckling in skin folds, scoliosis, and cutaneous neurofibromas, develop with age. Although most patients have a benign course, neurofibroma growth can accelerate during puberty or pregnancy, without spontaneous regression (Nguyen *et al.,* 2023).

Retroperitoneal manifestations are rare but significant in NF1. According to studies, their incidence ranges from 1% to 3% in NF1 patients. These manifestations include neurofibromatous masses that can compress adjacent structures, leading to varied symptoms such as abdominal pain, digestive disturbances, or urinary issues. Their identification is crucial for guiding diagnosis and management (Basile *et al.,* 2010).

CONCLUSION

Type 1 neurofibromatosis (NF1) is a genetic disorder characterized by significant clinical variability. Plexiform neurofibromas, including retroperitoneal ones, are rare but represent an important manifestation. Although often asymptomatic, NF1 requires regular monitoring due to the risk of tumor growth and associated complications.

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