

Neurofibromatosis type 1 (NF1): Experience of the Dermatology Department at Mohammed VI University Hospital in Oujda, Morocco

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Sir,

Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder caused by mutations in the NF1 gene located on chromosome 17, which encodes neurofibromin, a protein regulating cell growth [1,2].

This multisystem condition is characterized by cutaneous abnormalities (café-au-lait spots, neurofibromas), bone disorders (dysplasias, scoliosis), and neurological involvement (optic pathway gliomas, cognitive impairments) [3,4]. Diagnosis is based on clinical criteria, with skin manifestations often serving as the earliest indicators [3].

Dermatologists play a key role in the early detection and management of systemic complications [2,3,6].

The objective of this study was to evaluate the prevalence of complications associated with NF1 and to describe the epidemiological, clinical, and paraclinical characteristics of affected patients with the aim of improving multidisciplinary management and follow-up of this complex condition.

We conducted a retrospective, descriptive study over a ten-year period (June 2014–June 2024) at the Mohammed VI University Hospital in Oujda, including all patients diagnosed and followed for NF1 at our department.

A total of 28 patients were included, with a mean age of 14 years (range: 5–53 years). Among them, 17 were

female (60.71%) and 11 were male (39.29%). 46.4% had a first-degree family history of NF1, while 4.14% had second-degree relatives with the condition.

Regarding medical history, 21.4% of the patients had learning difficulties and a history of academic failure. Neurological manifestations were reported in 4% of the patients, with headaches being the most common symptom.

The main reason for consultation was the presence of hyperpigmented lesions, observed in 82.14% of the patients. Café-au-lait spots and lentigines were present in all cases. Crowe's sign was positive in 35% of the patients, and cutaneous or subcutaneous neurofibromas were observed in 67.8%. Plexiform neurofibromas were identified in 6 patients (21.4%).

As for paraclinical investigations, Lisch nodules were detected during the ophthalmologic examination in 9 patients (32.14%). Bone involvement was noted in 3 patients (10.71%), and 7 patients (25%) had lymphadenopathy on ultrasound.

All patients underwent brain MRI. Nine (32.14%) showed unidentified bright objects (UBOs), and 3 had intracranial tumors, one of which resulted in bulbo-medullary compression. These patients were referred to neurosurgery for further management.

Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder with an estimated prevalence of 1 in 2,500 to 3,000 births [2]. Clinical manifestations

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are diverse, involving the skin, nervous system, and bones, consistent with our observations. Café-au-lait spots, present in all patients, as well as lentigines and Crowe's sign (35%), align with existing literature on the cutaneous manifestations of NF1 [1].

Cutaneous and subcutaneous neurofibromas were seen in 67.8% of the cases, which was in line with reported data [3]. Plexiform neurofibromas were present in 21.4% of the cases, which is lower than rates described in the literature (up to 50%) [2]. Neurologically, our findings supported the high prevalence of low-grade gliomas among young adults with NF1 [2].

The presence of Lisch nodules (32.14%) and bone abnormalities (10.71%) in our cohort was consistent with literature-reported rates, highlighting the importance of ophthalmologic screening and radiological evaluation of skeletal anomalies in these patients [3,5].

The identification of space-occupying lesions requiring neurosurgical intervention emphasizes the critical need for multidisciplinary surveillance [2,5].

Despite the limitation of a small sample size, our findings underscored the importance of close follow-up for the early detection of tumor and skeletal complications and to optimize patient outcomes [6].

The dermatologist plays a crucial role in the early detection of NF1 manifestations and in identifying patients at risk of developing severe tumor or skeletal complications. Multidisciplinary monitoring remains essential to ensure optimal management of this complex disease [1,2].

Statement of Human and Animal Rights

All the procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the 2008 revision of the Declaration of Helsinki of 1975.

Statement of Informed Consent

Informed consent for participation in this study was obtained from all patients.

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