BEYOND NEUROFIBROMAS – RECOGNIZING LICHEN PLANUS IN A PATIENT WITH NEUROFIBROMATOSIS TYPE 1

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Summary

Introduction; Neurofibromatosis type 1 (NF1) is a common autosomal dominant disorder characterized by cutaneous neurofibromas, café-au-lait spots, and potential systemic involvement. Patients with NF1 may also develop unrelated dermatological conditions, which can pose diagnostic challenges. In this report, we describe the case of a patient with a known history of NF1 who presented with new inflammatory skin lesions, highlighting the importance of differential diagnosis in patients with pre-existing complex skin disorders.

Case presentation: We present the case of a 51-year-old male patient, previously diagnosed with neurofibromatosis type 1, who was admitted to our dermatology clinic for evaluation of erythematous, violaceous, scaly plaques located bilaterally in the pretibial region. The patient reported the recent onset of pruritic lesions without systemic symptoms. Clinical examination and laboratory investigations, including inflammatory markers and liver function tests, were performed. A clinical diagnosis of pretibial lichen planus was established based on the morphology and distribution of the lesions. No signs of neurofibromatosis-related complications were identified during hospitalization. The patient received topical corticosteroid therapy and antihistaminic receptor antagonists, with favorable evolution, and was advised regular dermatological follow-up.

Conclusions: In patients with pre-existing complex dermatological conditions such as NF1, the development of new skin lesions necessitates thorough clinical evaluation to distinguish between disease-related manifestations and unrelated dermatological entities. Early recognition and appropriate treatment of conditions like lichen planus can significantly improve patient outcomes.

Key words: *Neurofibromatosis type 1, lichen planus, pretibial lesions, differential diagnosis.*

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Introduction

Neurofibromatosis type 1 (NF1) is one of the most common autosomal dominant genodermatoses, with an estimated incidence of 1 in 3,000 individuals worldwide. The condition results from pathogenic variants in the NF1 gene on chromosome 17, which encodes neurofibromin, a tumor suppressor protein involved in the down-regulation of the RAS-MAPK signaling pathway; its deficiency predisposes patients to tumorigenesis, manifesting particularly across the skin and nervous system. (1)

From a dermatological perspective, NF1 is particularly significant, as cutaneous signs often provide the first clues to diagnosis and remain the most visible manifestations throughout life. The hallmark features include café-au-lait

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macules, which usually appear in early childhood, axillary and inguinal freckling, and multiple cutaneous neurofibromas that gradually increase in size and number with age. Neurofibromas are benign tumors of peripheral nerve sheath origin and may present as soft, flesh-colored papules or nodules distributed diffusely across the skin. Although generally asymptomatic, these lesions can cause cosmetic disfigurement, itching, or tenderness, and in some cases may lead to functional impairment depending on their anatomical location. Plexiformneurofibromas, though less common, carry a risk of malignant transformation into malignant peripheral nerve sheath tumors, making regular dermatological surveillance essential (1,2,8).

Beyond skin tumors, NF1's dermatologic spectrum also includes juvenile xanthogranulomas and nevus anemicus, with juvenile xanthogranulomas being quite frequent in pediatric NF1 and potentially predictive in uncertain diagnoses. Contemporary reviews highlight that cutaneous manifestations – not just cosmetic burdens – are often among the most distressing symptoms for NF1 patients and remain a therapeutic challenge. (1,8)

Despite the strong dermatological signature of NF1, it is crucial to recognize that patients may also develop cutaneous conditions unrelated to their genetic disease. This overlap presents a diagnostic challenge, as new lesions can easily be misinterpreted as part of the neurofibromatosis spectrum.

One such condition is lichen planus, an idiopathic, immune-mediated inflammatory dermatosis that affects both skin and mucous membranes – stratified squamous epithelia. Lichen planus typically manifests as violaceous, flat-topped, polygonal papules or plaques, often accompanied by the characteristic fine whitish reticulations of Wickham striae on their surface. The lesions are commonly distributed on the flexural surfaces of the wrists and forearms, the lumbar region, the pretibial area, and the oral or genital mucosa. Pruritus is a prominent and distressing symptom, frequently prompting medical consultation. (3,6)

Histologically, lichen planus is characterized by a band-like lymphocytic infiltrate at the dermo-epidermal junction and basal cell vacuolar degeneration, and sometimes colloid bodies, with direct immunofluorescence showing immunoglobulin or complement deposition; all of which reflecting its immune-mediated pathogenesis. While the exact trigger remains elusive, factors such as viral infections, medications, and stress have been implicated.(3,6,7)

Management typically begins with topical or systemic corticosteroids, with immunosuppressants as steroid sparing, and newer treatments such as JAK inhibitors or biologics targeting IL-12/23 or IL-17 emerging as promising options. (3,4)

The coexistence of NF1 and lichen planus illustrates the complexity of dermatological care in patients with pre-existing genodermatoses. When new erythematous, violaceous, or scaly plaques develop in an individual with NF1, clinicians must carefully consider whether the lesions represent an atypical presentation of neurofibromas, a secondary complication such as malignant transformation, or an entirely unrelated dermatosis such as lichen planus, psoriasis, or eczema. This distinction is critical, as the management strategies differ substantially. Whereas NF1-related cutaneous findings are generally managed through monitoring and, when necessary, surgical excision, lichen planus often responds well to anti-inflammatory approaches such as topical corticosteroids, calcineurin inhibitors, systemic immunomodulatory agents, or phototherapy, depending on severity and extent. (4,5)

Importantly, timely recognition and treatment of lichen planus in NF1 patients can significantly improve quality of life by reducing pruritus, preventing post-inflammatory hyperpigmentation, and minimizing the risk of chronicity. For dermatologists, this case scenario reinforces the importance of thorough clinical assessment, careful lesion morphology analysis, and, when appropriate, histopathological confirmation.

Ultimately, the overlap between a hereditary tumor predisposition disorder like NF1 and an acquired inflammatory condition like lichen planus highlights the need for individualized, vigilant dermatological care, ensuring that new findings are not overlooked or misattributed but are accurately diagnosed and managed in their own right.

Case presentation

We present the case of a 51-year-old male patient, previously diagnosed with neurofibromatosis type 1, who was admitted to our dermatology clinic for evaluation of erythematous, violaceous, scaly plaques located bilaterally in the pretibial region.

On examination, we noted the patient was of relatively short stature and presented the classical triad of neurofibromatosis - café-au-lait macules, axillary or inguinal freckling, and cutaneous neurofibromas – as established in the NIH diagnostic criteria. Multiple café-au-lait spots were distributed on the anterior and posterior aspects of the trunk, multiple disseminated neurofibromas on the face, trunk, abdomen and limbs and freckling under the arm, in the axillary region and also on the abdomen (Fig. 1). Multiple plexiformneurofibromas were observed on the posterior aspect of the trunk (Fig. 2).

Ophtalmological examination revealed Lisch nodules and visual deficit (Fig. 3).

The patient reported the recent onset of pruritic lesions without systemic symptoms. Clinical examination and laboratory investigations, including inflammatory markers and liver function tests, were performed and returned normal.

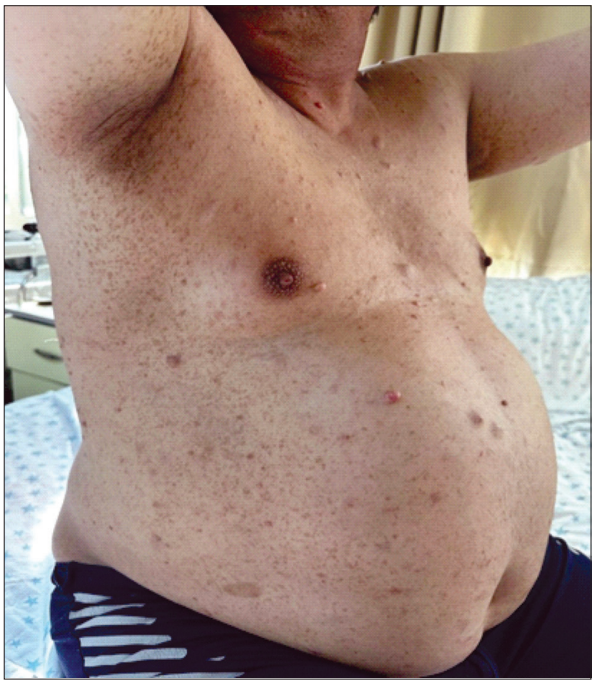

Figure 1 – Freckling, café-au-lait spots and multiple neurofibromas in the axillary region and anterior aspect of the trunk.

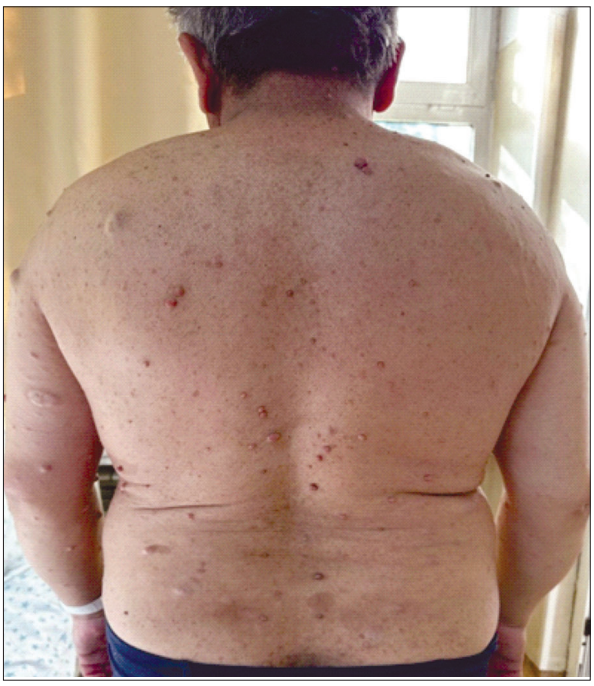

Figure 2 – Plexiformneurofibromas and café-au-lait spots on the posterior aspect of the trunk.

Pretibially, the patient presented bilateral shiny, erythematous, violaceous, dome-shaped

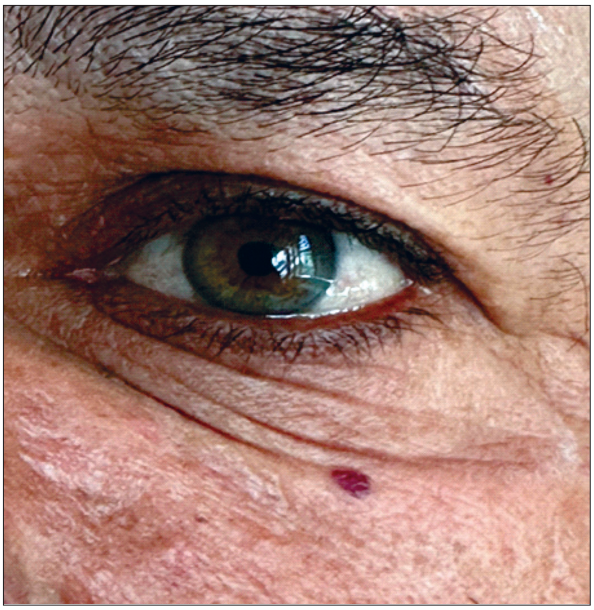

Figure 3 – Lisch nodules on ophtalmological examination.

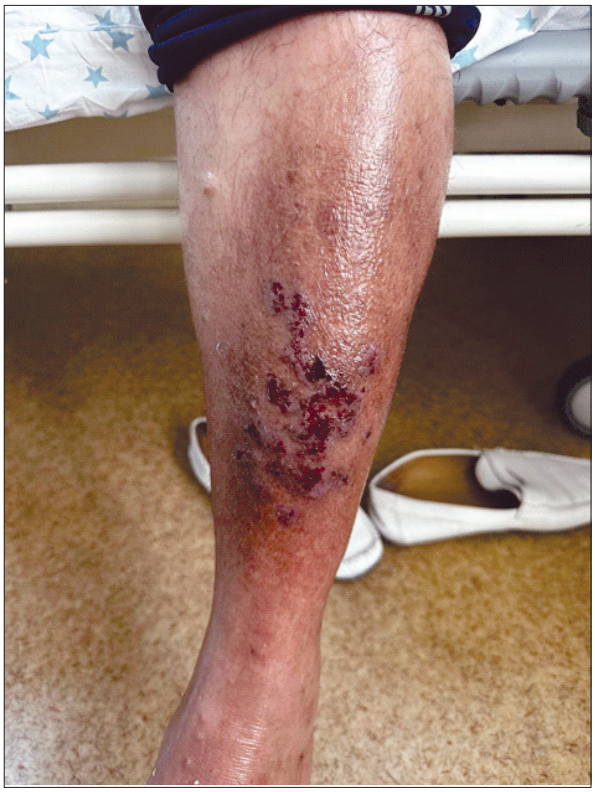

Figure 4 – Violaceous, dome shaped lesions clinically representative for pretibial lichen planus.

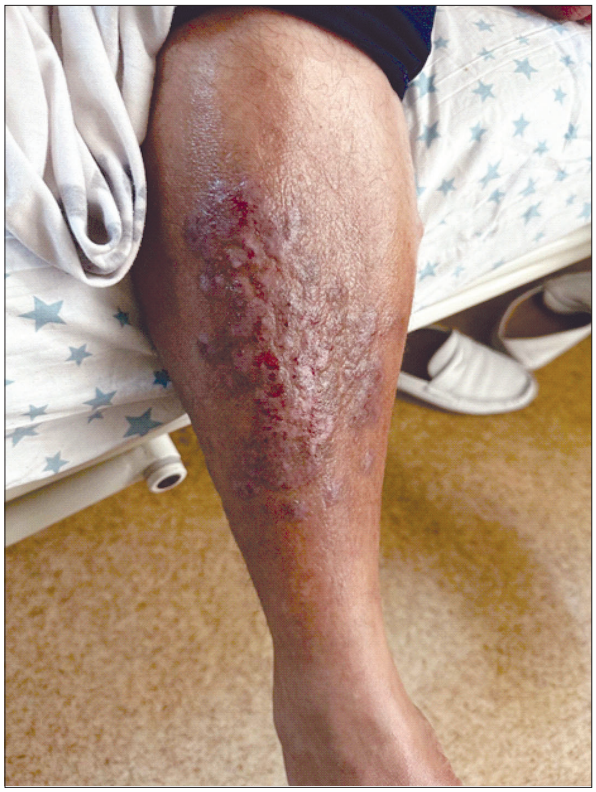

Figure 5 – Violaceous, dome shaped lesions clinically representative for pretibial lichen planus.

scaly plaques which were intensly pruritic (Fig 4, Fig. 5). The lesions has been present for multiple months and had previously been mistakenly attributed to NF1 by the patient and primary care providers.

A clinical diagnosis of pretibial Lichen Planus was established based on the morphology and distribution, after visual and dermatoscopic evaluation of mature looking lesions, which showed Wickham striae, vascular structures (multiple red dots and radial capillaries) and diffuse brownish coloration (Fig. 6).

Diagnosis was confirmed by histopathological examination of tissue biopsy from the pretibial area, which showed degeneration of the basal layer of the epidermis and a band like lymphocytic infiltrate obscuring the dermo-epidermal junction; as well as irregular epidermal hyperplasia forming a saw-tooth appearance with wedge-shaped hypergranulosis . The basal layer of the epidermis exhibited vacuolar de-

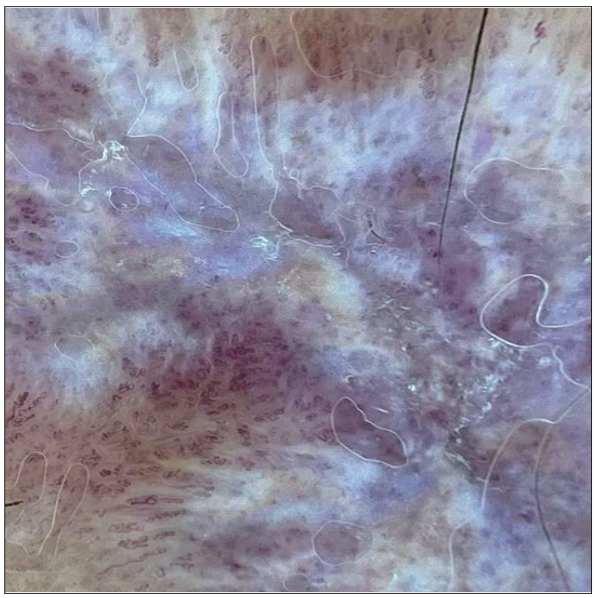

Figure 6 – Dermoscopy.

generation with typically prominent necrosis of individual keratinocytes. These findings were consistent with the diagnosis of lichen planus.

No signs of neurofibromatosis-related complications were identified during hospitalization.

The patient received topical corticosteroid therapy with highly potent corticosteroids under occlusive bandages and antihistaminic receptor antagonists, with favorable evolution, and was advised regular dermatological follow-up.

Discussions

Neurofibromatosis type 1 (NF1) is a genodermatosis with highly recognizable cutaneous manifestations, including café-au-lait macules, axillary or inguinal freckling, and multiple cutaneous neurofibromas. These features are central to both diagnosis and follow-up, as neurofibromas, while typically benign, may increase in number and size throughout life and occasionally undergo malignant transformation into malignant peripheral nerve sheath tumors (1,2). Given this strong dermatological signature, clinicians often attribute the emergence of new skin lesions in NF1 patients to the natural course of the disease. However, such diagnostic anchoring can obscure the identification of unrelated cutaneous disorders that require distinct management.

Lichen planus (LP) represents one such condition. It is an acquired, immune-mediated dermatosis characterized by violaceous, polygonal, flat-topped papules or plaques, often accompanied by Wickham striae and pruritus. The pretibial location observed in our patient is consistent with one of the well-documented distribution patterns of LP. Histopathologically, LP demonstrates a dense, band-like lymphocytic infiltrate at the dermo-epidermal junction with basal cell degeneration, reflecting its immune-driven pathogenesis (3,4). Unlike NF1, LP is not genetically determined but arises sporadically, with potential associations including viral infections, medications, and psychosocial stress.

The co-occurrence of NF1 and LP is striking, as it has rarely been described in the literature. While previous reports have noted associations between NF1 and other dermatological or autoimmune conditions, such as lichen sclerosus or juvenile xanthogranulomas, LP remains uncommon in this context (5). This rarity underscores the importance of thorough clinical evaluation when NF1 patients present with new inflammatory or erythematous lesions. Differential diagnoses should encompass atypical neurofibromas, malignant peripheral nerve sheath tumors, psoriasis, eczema, and inflammatory dermatoses such as LP.

Our case illustrates that unrelated inflammatory skin diseases in NF1 can be successfully managed with conventional therapy once accurately diagnosed. The favorable response to topical corticosteroids and antihistamines highlights the importance of timely recognition and treatment of LP, even in the context of com-plex genetic skin disorders.

Furthermore, this case reinforces the need for regular dermatological follow-up in NF1 not only to monitor for disease-related complications but also to detect unrelated cutaneous entities that may impact quality of life.

Conclusions

In patients with pre-existing complex dermatological conditions such as NF1, the development of new skin lesions necessitates thorough clinical evaluation to distinguish between disease-related manifestations and unrelated dermatological entities. Early recognition and appropriate treatment of conditions like lichen planus can significantly improve patient outcomes.

In summary, NF1 – and its expansive cutaneous phenotype – necessitates diligent surveillance, especially in the presence of new or evolving lesions. The possibility of concurrent inflammatory dermatoses, though rare, must be entertained. This approach ensures timely, condition – specific management, avoids misattribution to the underlying genodermatosis, and ultimately enhances personalized patient care in dermatology.

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Conflict of interest
NONE DECLARED

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