Dermatology Practical & Conceptual

Nevus Unius Lateris and Nevus Anemicus in a Patient With Neurofibromatosis Type 1: Noninvasive Imaging With Line-Field Confocal Optical Coherence Tomography and High-Resolution Video Dermoscopy

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Key words: Nevus Unius Lateris, Neurofibromatosis Type 1, LC-OCT, Nevus Anemicus, High-Resolution Video Dermoscopy

Citation: Falcinelli F, Calabrese L, Rubegni P, Tognetti L. Nevus Unius Lateris and Nevus Anemicus in a Patient With Neurofibromatosis Type 1: Noninvasive Imaging With Line-Field Confocal Optical Coherence Tomography and High-Resolution Video Dermoscopy. *Dermatol Pract Concept*. 2025;15(4):5633. DOI: https://doi.org/10.5826/dpc.1504a5633

Accepted: April 23, 2025; **Published:** October 2025

Funding: None.

Competing Interests: None.

Authorship: All authors have contributed significantly to this publication.

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Introduction

Neurofibromatosis type 1 (NF1) is a multisystem autosomal dominant disorder that leads to abnormalities in the nervous and skeletal system; cutaneous hallmarks for diagnosis include café-au-lait macules, skinfold freckling, Lisch nodules, and dermal neurofibromas [1]. Nevus anemicus (NA) is a congenital vascular malformation appearing as a hypopigmented, pale, well-defined oval patch. Due to a focal increased sensitivity of blood vessels to catecholamines, NA does not become erythematous after scratching, heat, or cold, while reactive erythema appears in the surrounding skin [2]. NA is considered a specific diagnostic clue for NF1 [1,2]. Nevus unius lateris (NUL) is an extremely rare congenital hamartoma characterized by papillomatous, verrucous to velvety papules/plaques distributed in a linear blaschkoid pattern and affecting one half of the body, regarded as a systematized verrucous variant of epidermal nevus [3].

Case Presentation

A 13-year-old male presented due to the appearance in the preceding few of years of hypopigmented patches localized at the lumbar, sternal, and pectoral areas (Figure 1). He was diagnosed with neurofibromatosis type 1 (NF1) during childhood and skinfolds freckling and café-au-lait patches were present. Multiple verrucous brownish lesions arranged along the Blaschko lines, limited to the left half of the trunk,

were also visible. The lesions had increased in dimension and number over time in the previous 10 years and had been diagnosed as warts. The patient had cognitive retardation and learning disorders. The parents denied their consent to perform skin biopsies.

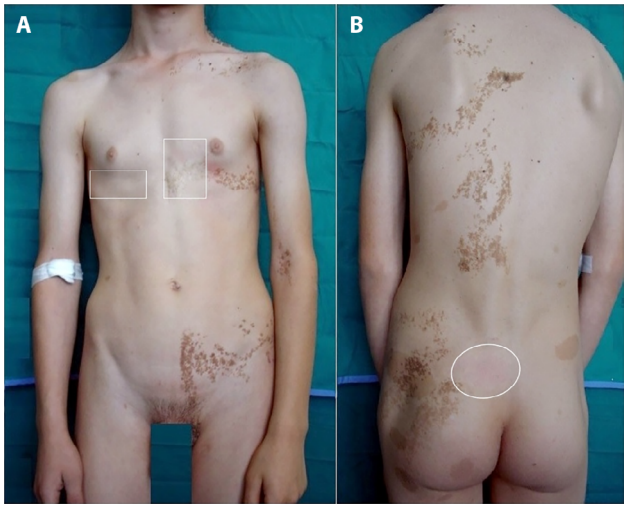

Figure 1. Patient's clinical appearance at presentation: many café-au-lait patches and macules were visible all over the trunk; hypopigmented patches are visible at lumbar (B, white circle), sternal, and pectoral areas (A, white squares), corresponding to nevus anemicus. Multiple verrucous brownish lesions arranged along the Blaschko lines and delimited bythe midline to the left half of the trunk corresponding to nevus unius lateris are also visible.

Noninvasive skin examination with line-field confocal optical coherence tomography (LC-OCT) and high-resolution videodermoscopy (HRVD) were performed on the verrucous lesions and on the hypopigmented patch, revealing hyperkeratosis and acanthosis in the former and normal epidermis with narrowed dermal vessels in the latter (Figure 2).

Conclusion

Based on clinical and imaging findings, a diagnosis of NULNA-NF1 association was posed.

HRVD (30x–100x magnification) and LC-OCT are two recently introduced new noninvasive imaging techniques which can provide high-resolution images of the skin on vertical and horizontal planes with cellular resolution, and they prove to be particularly helpful for doubtful lesions in the pediatric populations [4,5]. HRVD examination is rapid and offers more detailed information than standard dermoscopy (17x), both in contact and non-contact mode, and is very useful in the visualization of vascular structures [4]. LC-OCT takes both vertical/horizontal and 3D images in vivo in real time and to a depth of 500 µm and a FOV of 1.2 mm. The result is histological-like imaging of the explored skin area in gray-scale, like a "virtual biopsy" [4,5].

Dermoscopy of NA can highlight thinned blood vessels in the center, compensatory flare, and blending with the surrounding skin [4], while histology is usually normal [1,2].

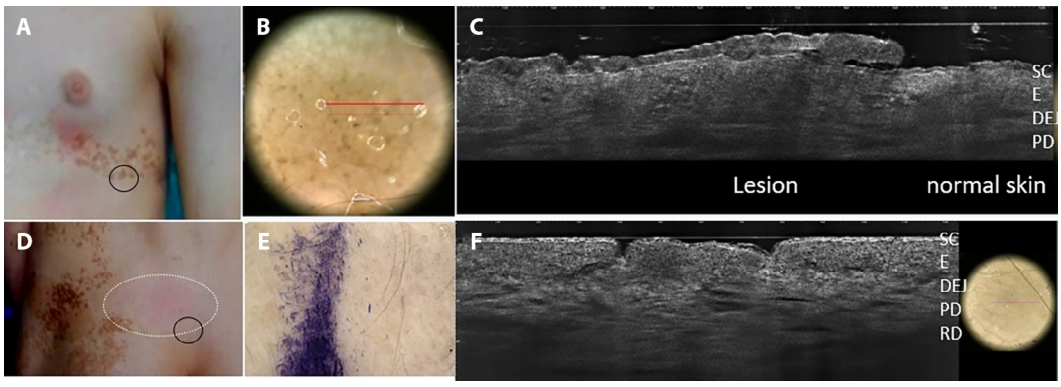

Figure 2. (A-C) Combined noninvasive imaging of nevus unius lateris performed at lesional margin on the submammary verrucous brownish lesions (A, black circle). (B) Polarized dermoscopy 20x highlights multiple brownish crusts and dots over a yellowish background. (C) corresponding vertical LC-OCT examination showed acanthosis with hyperkeratotic layer homogeneously distributed over the lesion surface. Dermal vessels appear as hyporeflective holes, normal in shape. (D-F) Combined noninvasive of lumbar nevus anemicus (white dotted circle) after moderate skin rubbing. (E) High-resolution video dermoscopy 30x and (F) polarized dermoscopy (20x) show both a pale hypopigmented skin appearance with signs of atrophy. (G) LC-OCT vertical examination taken at the lesional margin (D, black circle) identifies few thinned well-defined papillary vessels (v) and normal epidermis, with no sign of atrophy at the lesional site, while (F) normal-shaped vascular lacunae are seen in surrounding skin as hyporeflective dark holes. Abbreviations: SC, stratum corneum; E, epidermis; DEJ, dermal-epidermal junction; PD, papillary dermis; RD, reticular dermis; v, dermal vessels. Note: The red line inside the dermoscopic round frames corresponds to the exact point of LC-OCT examination (i.e., length of the LC-OCT frame).

Dermoscopic features of NUL include large brown circles on a brownish background; hyperkeratosis, acanthosis, and papillomatosis are detected by histopathology [3] as well as by LC-OCT [4,5], along with possible focal increase in basal melanin.

The combined HRVD and LC-OCT examination at multiple lesional sites oriented us toward the diagnostic suspicion of an NUL-NA-NF1 association. This high-resolution imaging also allowed us to respect the parents' wishes to avoid multiple skin biopsies with their consequent aesthetic impact and scarring in a fragile patient.

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