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Introduction

Spitz tumors are a heterogenous group of melanocytic neoplasms, which on histology demonstrate enlarged oval, epithelioid or spindled melanocytes, often in association with epidermal hyperplasia. The degree of pigmentation and presence or absence of maturation is variable. The molecular-genetic basis for these variations in histomorphology is increasingly being recognized. Spitz tumors typically harbor HRAS activating mutations, inactivation or deletion of BAP1 or gene fusions involving the kinases ROS1, ALK, NTRK1, BRAF, RET, MET or NTRK3 [1]. Some of these aberrations have been associated with progression to melanoma, while others have not [2].

There is little in the literature regarding the impact of particular molecular subtypes of Spitz on clinical-dermoscopic appearance. Most publications describe the clinical-dermoscopic features common to Spitz tumors as an entire category. We deduce that, as many of these neoplasms have characteristic histomorphology, clinical-dermoscopic features may be relatively consistent for a particular molecular-genetic aberration. Herein, we describe the clinical-dermoscopic features of a Spitz tumor with an LMNA-NTRK1 fusion presenting in an adult.

Case Presentation

A 51-year-old man with a history of numerous clinically atypical pigmented lesions, multiple biopsy-confirmed dysplastic nevi, and a first-degree relative with metastatic melanoma presented for a follow-up comprehensive skin examination. Due to the patient’s high risk, his skin surface was being surveyed longitudinally with digital whole-body photography and high-resolution dermoscopy. A new lesion was identified on his left calf, which was not present on exam 6 months prior.

Clinical examination revealed a smooth, red, dome-shaped 5 × 5 mm papule with little surface change (Figure 1A). On dermoscopy, the lesion was symmetric and demonstrated a regular array of coiled-glomerular vessels admixed with crystalline structures (Figure 1B). On histopathology, there was a broad compound proliferation of enlarged spindled and epithelioid (amelanotic) melanocytes with associated epidermal hyperplasia (Figures 2 and 3). Many junctional melanocytic nests demonstrating peripheral clefting and occasional Kamino bodies were observed. Thin and elongated “filigree-like” rete ridges were seen, occasionally extending into the superficial tumor aggregates. In the dermis, there were “lobulated nests” along with conspicuous melanocytic mat-
The presence of a dome-shaped red papule with a regular array of glomeruloid vessels admixed with crystalline structures should prompt clinical suspicion of a Spitz tumor with a \( NTRK1 \) fusion. Although some variants of Bowen disease contain similar vessel morphology, the coexistence of crystalline structures and lack of scale would be unusual. Furthermore, the diffuse (as opposed to grouped) arrangement of the vessels is not typical for Bowen disease. Additional larger series are needed to replicate these findings and inform on the clinical-dermoscopic features of particular molecularly defined Spitz tumor subtypes.

References