

Amelanotic Melanoma in Oculocutaneous Albinism: Clinical Presentation and Diagnostic Pitfalls with Dermoscopic-Histopathological Correlation

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Introduction

Oculocutaneous albinism (OCA) comprises a group of rare autosomal disorders of pigmentation caused by recessive mutations in genes involved in melanin biosynthesis, with a prevalence of 1:17.000 cases [1]. OCA presents with heterogeneous phenotypes ranging from a partial to a complete lack of pigmentation. Clinically, OCA-Ia can be distinguished by the total absence of melanin, while other types (OCA-Ib to OCA-VIII) show minimal pigmentation. This melanin reduction increases the risk of UV-induced skin cancers,

particularly nonmelanoma skin cancer (NMSC), with melanomas being rare (~1% of cases) and typically amelanotic. We report clinical, dermoscopic, and histopathological features of a case of amelanotic melanoma (AM) in a patient with OCA.

Case Presentation

A 28-year-old Iranian man with OCA, fair hair, gray-blue eyes, and bilateral nystagmus presented with a long-standing reddish pink plaque on his back prone to bleeding. The

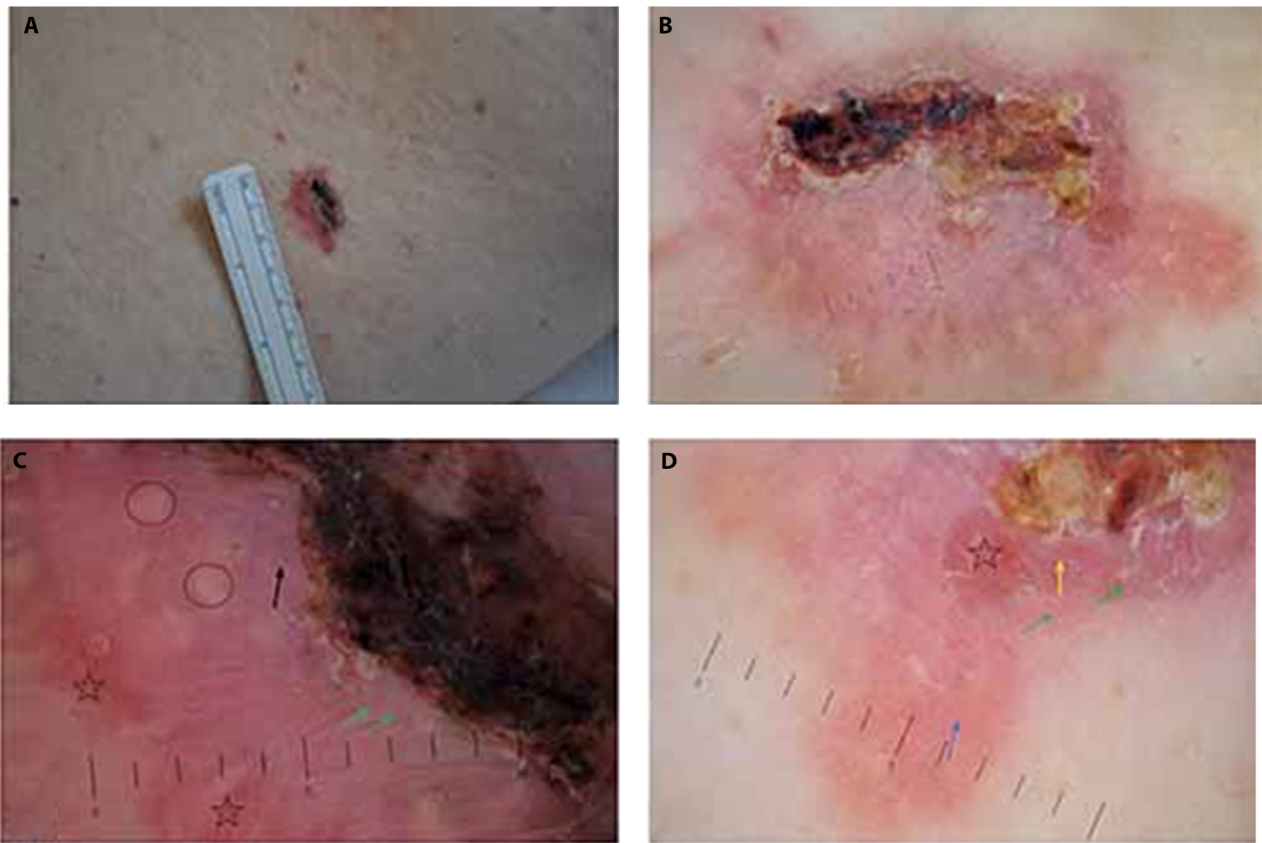


Figure 1. Clinical and dermoscopic images of an ulcerated 1.8-mm thick superficial spreading melanoma on the back of a 28-year-old man with oculocutaneous albinism. (A) In the clinical image an asymmetric reddish-pink ulcerated plaque partially covered by a dark crust can be observed. (B) In the dermoscopic image of the same melanoma milky red areas, scales, shiny white streaks, unfocused roundish whitish-yellow areas, and a polymorphous vascular pattern can be seen. The right part of the lesion had a reddish-yellow-orange hemorrhagic crust from ulceration. (C) Magnified details of milky red areas (star), milky red globules (black arrow), multiple unfocused roundish whitish-yellow areas (circles), hairpin vessels (green arrow), scales, and black hemorrhagic crust in the right part of the lesion. (D) Magnified details of polymorphous vascular pattern consisting of dotted (blue arrow), linear irregular (yellow arrow), hairpin (green arrows), and milky red areas (star). Scales and yellow-orange hemorrhagic crust can also be observed in the upper right part of the lesion.

lesion was asymmetric, ulcerated, and measured 3 cm in diameter, with a yellowish-brown crust (Figure 1A). Dermoscopy revealed a structureless pattern including scales, shiny white structures, unfocused whitish-yellow and milky red areas, a reddish-orange hemorrhagic crust, and a polymorphous vascular pattern consisting of dotted, hairpin, and linear irregular vessels (Figure 1B–D). The lesion was surgically excised, revealing a superficial spreading invasive melanoma with a Breslow thickness of 1.8 mm, a mitotic index of 8/mm², no vascular invasion, and a lymphohistiocytic infiltration (Figure 2). Interestingly, hyperplastic sebaceous glands in the upper part of the reticular dermis that were almost attached to the epidermis could be clearly identified (Figure 2C and D).

Conclusion

Melanoma in OCA patients is extremely rare, with only 56 reported cases, 33 of which were AM [1]. Few reports [1-3] have described the dermoscopic features of AM in

OCA, which include white shiny streaks, yellowish areas, and polymorphous vascular patterns, consistent with our findings. Atypical vascular patterns, such as polymorphic vessels and milky red areas, are characteristic of amelanotic/hypomelanotic melanoma [4,5], with vessel polymorphism increasing with melanoma thickness. Our case also showed unfocused roundish whitish-yellow areas, typically seen in benign lesions like dermal nevi or seborrheic keratosis, where they correspond to intraepidermal keratin-filled cysts. Here they histologically corresponded to hyperplastic sebaceous glands, an association not previously reported in melanomas. In AM patients with OCA, yellowish areas have already been described, but a possible histopathologic substrate has never been suggested [2,3]. Sebaceous glands have been found to contain melanocytes, potentially serving as source of melanocyte stem cells [6]. These melanocytes might produce cytokines, catecholamines, nitric oxide, and pro-opiomelanocortin peptide in response to ultraviolet irradiation and other stimuli, affecting sebocytes [6]. Their role in melanoma remains unclear and warrants further research.

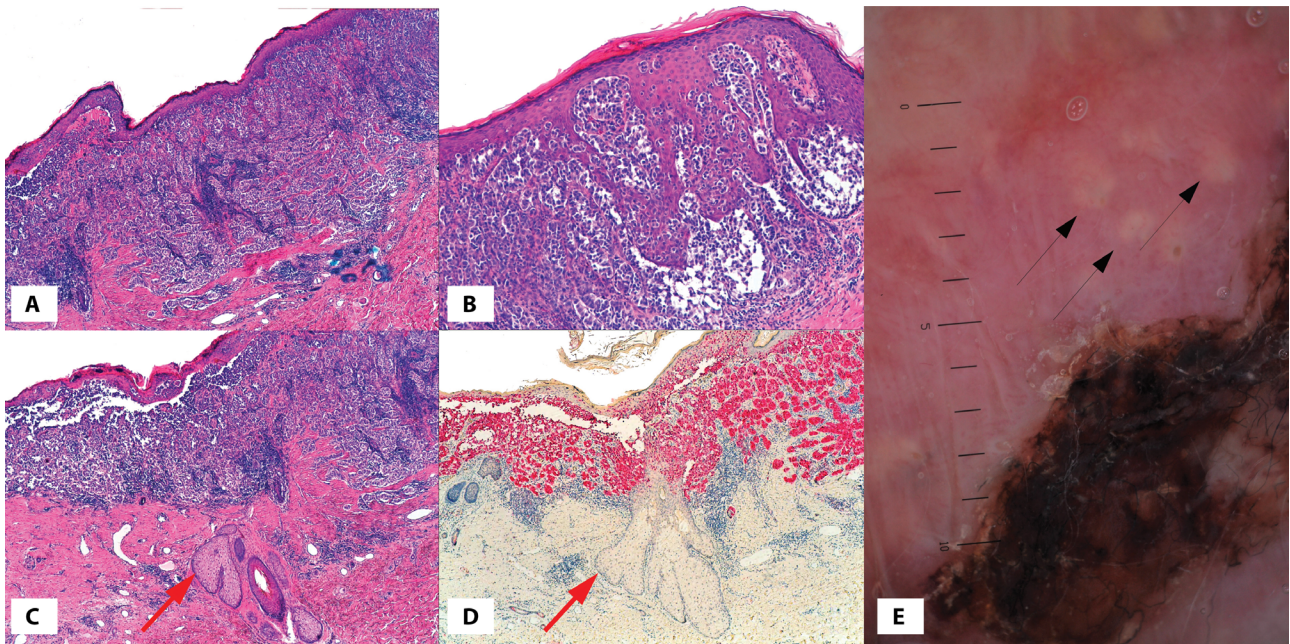


Figure 2. (A) Atypical melanocytes in irregular nests and single units in the epidermis and sheets of atypical melanocytes in the dermis with lymphohistiocytic infiltrate (H&E, $\times 20$). (B) Atypical melanocytes in irregular nests and single units in all layers of the epidermis (H&E, $\times 200$). (C) Atypical melanocytes in irregular nests and single units in the epidermis and sheets of atypical melanocytes in the dermis with lymphohistiocytic infiltrate. interestingly, in the upper part of the reticular dermis, a hair follicle with sebaceous glands is present (red arrow) (H&E, $\times 20$). (D) Melan-A staining highlights the atypical melanocytes in irregular nests and single units in the epidermis and the sheets of atypical melanocytes in the dermis. Hyperplastic sebaceous glands almost attached to the epidermis are clearly observed (red arrow) (H&E, $\times 20$). (E) Dermoscopic-pathologic correlation: the unfocused roundish whitish-yellow areas (black arrows) might well correlate with the hyperplastic sebaceous glands described in C and D.

In OCA patients, the presence of a pink lesion should raise suspicion for both NMSC and AM. Dermoscopy can aid in distinguishing AM from other skin cancers, but regular clinical-dermoscopic examinations every 6-12 months are recommended. For challenging lesions, digital follow-up and reflectance confocal microscopy should complement total body dermoscopy. Dermoscopy, through the identification of polymorphic vessels, shiny white streaks, and whitish-yellow areas, can enhance early detection of AM. The dermoscopic-pathological correlation of whitish-yellow areas, especially in OCA patients, requires further investigation.

References

1. Ruiz-Sanchez D, Garabito Solovera E, Valtueña J, et al. Amelanotic melanoma in a patient with oculocutaneous albinism. *Dermatol Online J.* 2020;26:1–3 DOI: 10.5070/D3265048785 PMID: 32621707
2. De Luca DA, Bollea Garlatti LA, Galimberti GN, Galimberti RL. Amelanotic melanoma in albinism: the power of dermoscopy. *J Eur Acad Dermatol Venereol.* 2016;30:1390–1394 DOI: 10.1111/jdv.13264 PMID: 26290313
3. Uyar B, Faruk Elmas O, Kilitci A, Tad M. Dermoscopy of amelanotic melanoma in a patient with oculocutaneous albinism. *Dermatol Pract Concept.* 2020;10:1–2 DOI: 10.1111/jdv.13264 PMID: 26290313
4. Zalaudek I, Kreusch J, Giacomel J, Ferrara G, Catricalà C, Argenziano G. How to diagnose nonpigmented skin tumors: a review of vascular structures seen with dermoscopy. *J Am Acad Dermatol.* 2010;63:361–374 DOI: 10.1016/j.jaad.2009.11.698 PMID: 20708469
5. Pizzichetta MA, Talamini R, Stanganelli I, et al. Amelanotic/hypomelanotic: clinical and dermoscopic features. *Br J Dermatol.* 2004;150:1117–1124 DOI: 10.1111/j.1365-2133.2004.05928.x PMID: 15214897
6. Jang YH, Kim SL, Lee JS, et al. Possible existence of melanocytes or melanoblasts in human sebaceous glands. *Ann Dermatol.* 2014;26:469–473 DOI: 10.5021/ad.2014.26.4.469 PMID: 25143675