Other Faces of Darier Disease

Otras caras de la enfermedad de Darier

Darier disease—or, more appropriately, Darier-White disease—will perhaps be one of the few eponymous disease names that will survive in the dermatology of the future. The eponym’s survival will surely be assisted by the clear inappropriateness of the alternative names that have been proposed: keratosis follicularis and, more recently, dyskeratosis follicularis.

The last 2 decades have yielded key molecular and genetic knowledge. The relationship between Darier disease and the ATP2A2 gene and its chromosomal locus was clearly established, and the aura of mystery surrounding this genodermatosis seemed to vanish. But nothing could be further from reality: the wealth of expression and the clinical and therapeutic unpredictability of Darier disease continue to fascinate us.

The cases reported in this issue by Flores-Terry et al.¹ provide a lovely example of the diversity of Darier disease. They show that, in parallel to—and even in spite of—advances in basic research, clinical practice continues to attract and captivate us on a daily basis. In fact, suspicion of Darier disease based on those peculiar hemorrhagic blisters at acral sites is a good example of the old maxim, “If you don’t think of it, you won’t diagnose it,” and it shows us the way to clinical excellence in dermatology.

Reference


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