WHAT IS THE DIAGNOSIS

Serpiginous Traces in Dermatology: Case Report of Elastosis Perforans Serpiginosa in a Patient with Down Syndrome

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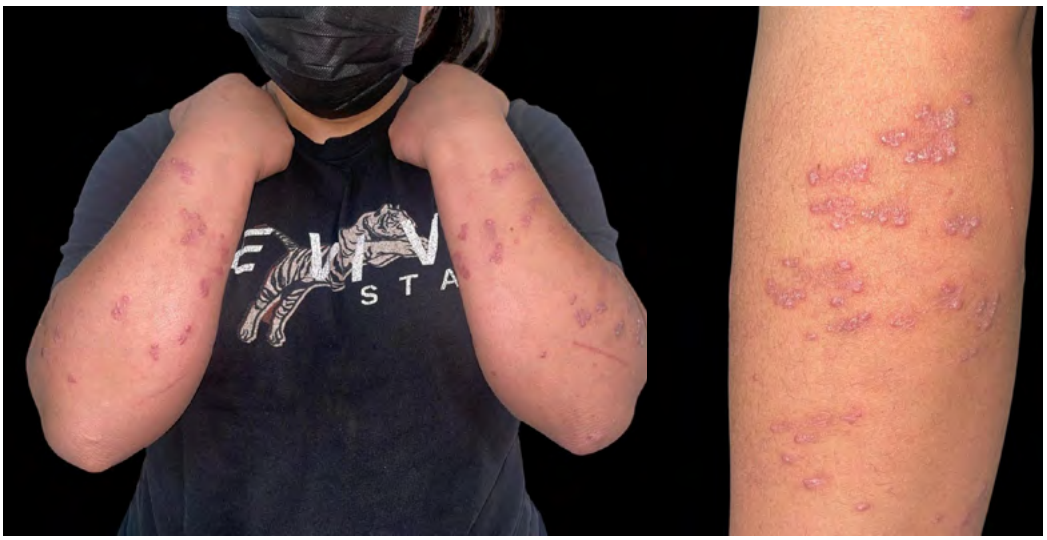
Key words: elastosis perforans serpiginosa, Down syndrome, perforating dermatosis

Date of receipt: 09/11/2024
Date of acceptance: 01/04/2025

How to cite this article: Félix C., López L., Santofimio M., Aguilar Y., Pacheco R. Trazos Serpiginosos en la Dermatología: Haga su diagnóstico. Rev Dermatol Cent Úraga. 2025;7(2).

WHAT'S YOUR DIAGNOSIS?

We present the case of a 25-year-old female patient with a personal history of Down syndrome. She presented cutaneous lesions on the forearms for two years. They appeared without apparent cause and were accompanied by intense pruritus. She had previously been treated with emollients and topical steroids without improvement. On physical examination, she showed a localized dermatosis on the dorsal side of both forearms, characterized by grouped erythematous-violaceous hyperkeratotic papules in a pseudo-annular pattern (Figures 1 and 2)



Figures 1 and 2. Hyperkeratotic erythematous-violaceous pseudo-annular papules on the dorsal forearms.

WHAT IS YOUR DIAGNOSIS?

A skin biopsy was performed, showing irregular acanthosis, hyperkeratosis with parakeratosis, and dilated follicles through which collagen and elastic tissue extrude. A keratin plug was seen within an epidermal invagination, surrounded by acanthosis and hyperkeratosis (Figure 3A), and the Verhoeff-van Gieson stain revealed transepidermal elimination of elastic fibers in black, findings consistent with a perforating dermatosis. Based on these findings, clinical characteristics, and the patient's history of Down syndrome, the case was diagnosed as elastosis perforans serpiginosa. Treatment with 0.1% topical retinoic acid was initiated while awaiting results.

DISCUSSION

Elastosis perforans serpiginosa (EPS) is a rare primary perforating dermatosis. It affects men more frequently than women and typically appears in early adulthood.[1] The most commonly affected areas are the neck, face, arms, and skin folds.[2] EPS is associated with genetic disorders such as Down syndrome, Ehlers-Danlos syndrome, osteogenesis imperfecta, Marfan syndrome, pseudoxanthoma elasticum, Rothmund-Thomson syndrome, and acrogeria.[3]

The condition arises from an overproduction of morphologically and biochemically altered elastic fibers, often accompanied by abnormal collagen fibers. These altered elastic fibers, perceived as foreign material, are eliminated through transepidermal channels.[4] It has been suggested that impaired phagocytic function in these patients may promote epidermal elimination as an alternative pathway for cellular debris disposal.[5]

EPS typically presents as small keratotic papules arranged in an annular or serpiginous pattern, and lesions may be asymptomatic or pruritic. Differential diagnoses include annular granuloma, sarcoidosis, cutaneous calcinosis, and tinea corporis.[6]

Histopathological findings include increased thickened elastic fibers in the papillary dermis. These are extruded through channels filled with fragmented elastic fibers and basophilic nuclear debris. As they enter the channel—typically capped by a keratin plug—the fibers lose their staining properties. The epidermis becomes hyperplastic and acanthotic, with a claw-like appearance as it attempts to expel the altered elastic fibers.[7]

Treating EPS is challenging. Topical steroids are commonly used, but their effectiveness is limited. Treatment options include topical retinoids, tacrolimus, CO2 laser therapy, cryotherapy, and narrowband UVB phototherapy. These therapies have shown variable success in reducing lesions and relieving pruritus. As EPS tends to be chronic and treatment-resistant, an individualized therapeutic approach and regular follow-up are essential to improve patient quality of life.

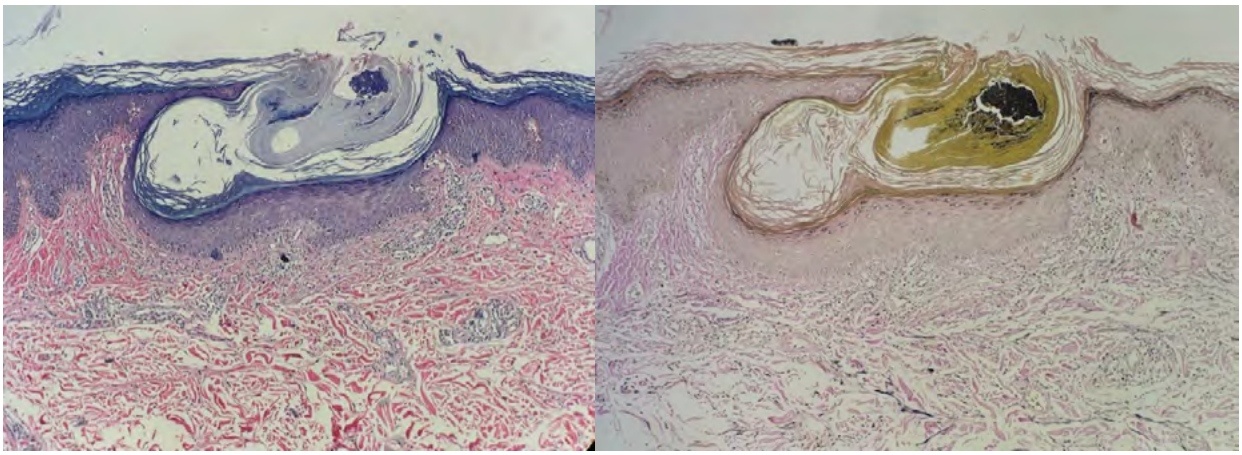


Figure 3. A. Keratin plug within an epidermal invagination surrounded by acanthosis and hyperkeratosis. B. Transepidermal elimination of elastic fibers (black) (Verhoeff-van Gieson stain).

In conclusion, elastosis perforans serpiginosa is a rare disorder often associated with genetic conditions such as Down syndrome. Its clinical presentation—characterized by hyperkeratotic papules in a serpiginous pattern—and histopathological features of transepithelial elimination of elastic fibers are crucial for diagnosis. Despite multiple available treatments, EPS often shows resistance to conventional therapies, highlighting the need for individualized and multidisciplinary management.

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