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Photo vignette

Periumbilical perforating pseudoxanthoma elasticum

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Abstract

A 50-year-old woman presented with a 2-year history of a yellowish plaque studded with red brown keratotic papules in the periumbilical region. Histopathological examination from the yellow plaque showed curled and granular elastic fibers in the mid and lower dermis. Histopathological examination from a keratotic papule showed pathological elastic fibers and dense chronic inflammatory cells around areas of perforation. Clinicopathological correlation established periumbilical perforating pseudoxanthoma elasticum as the final diagnosis.

Keywords: Periumbilical perforating pseudoxanthoma elasticum, pseudoxanthoma elasticum, fragmented elastic fibers

Case synopsis

A 50-year-old, gravida 7 (all born by normal vaginal delivery), obese woman presented with an asymptomatic plaque on the abdomen of 2 years duration. New lesions kept appearing forming a confluent plaque. Over time red-brown colored papules with central keratotic material developed over this plaque. Her medical history was notable for hypertension. Her family history was unremarkable. On examination, a reticulated yellowish plaque was noted in the periumbilical region. The surface of this plaque was studded with red-brown papules and keratotic papules (Figure 1).

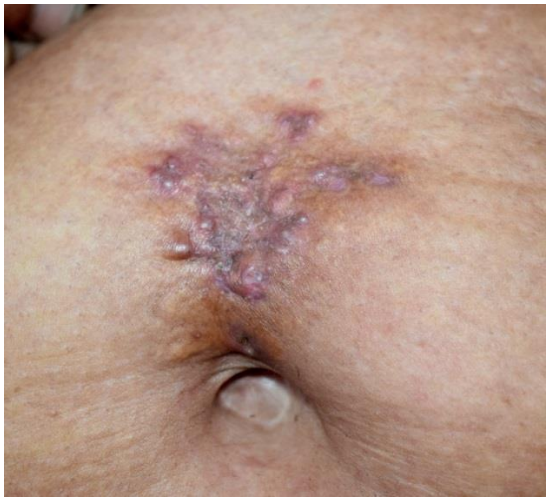

Figure 1. Red-brown, keratotic, periumbilical papules forming a plaque.

Results of routine laboratory tests were normal. Abdominal ultrasonography, echocardiography, and fundus examination were found to be normal. Two punch biopsy specimens were taken of a yellowish flat papule and a keratotic papule. The former showed curled and granular elastic fibers in the mid and lower dermis, consistent with pseudoxanthoma elasticum (Figure 2)

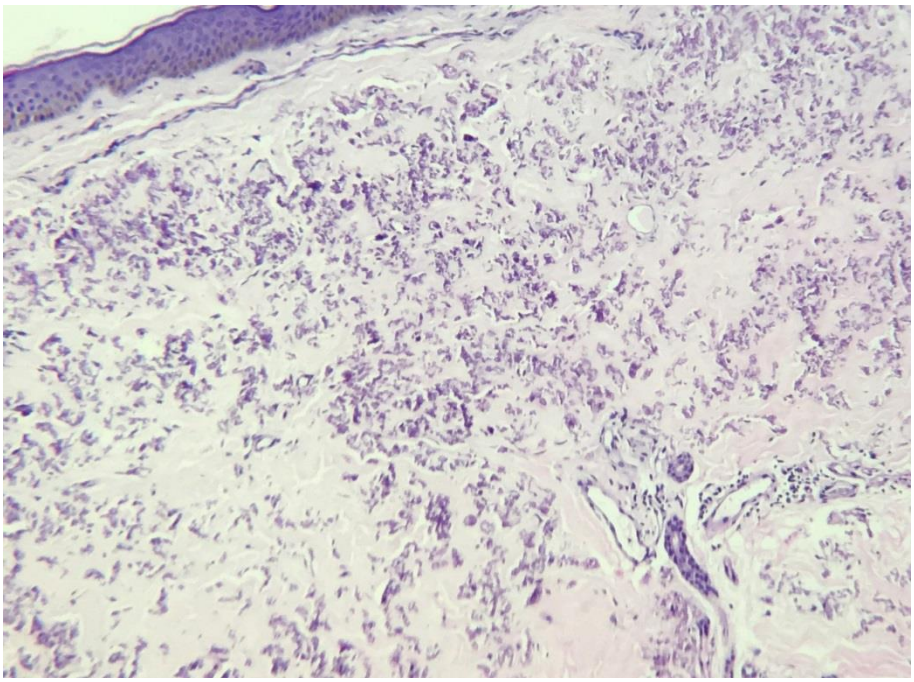

Figure 2. Curled and granular elastic fibers. H&E, 100x.

and the latter showed pathological elastic fibers and dense chronic inflammatory cells around a perforation, consistent with perforating pseudoxanthoma elasticum (Figure 3).

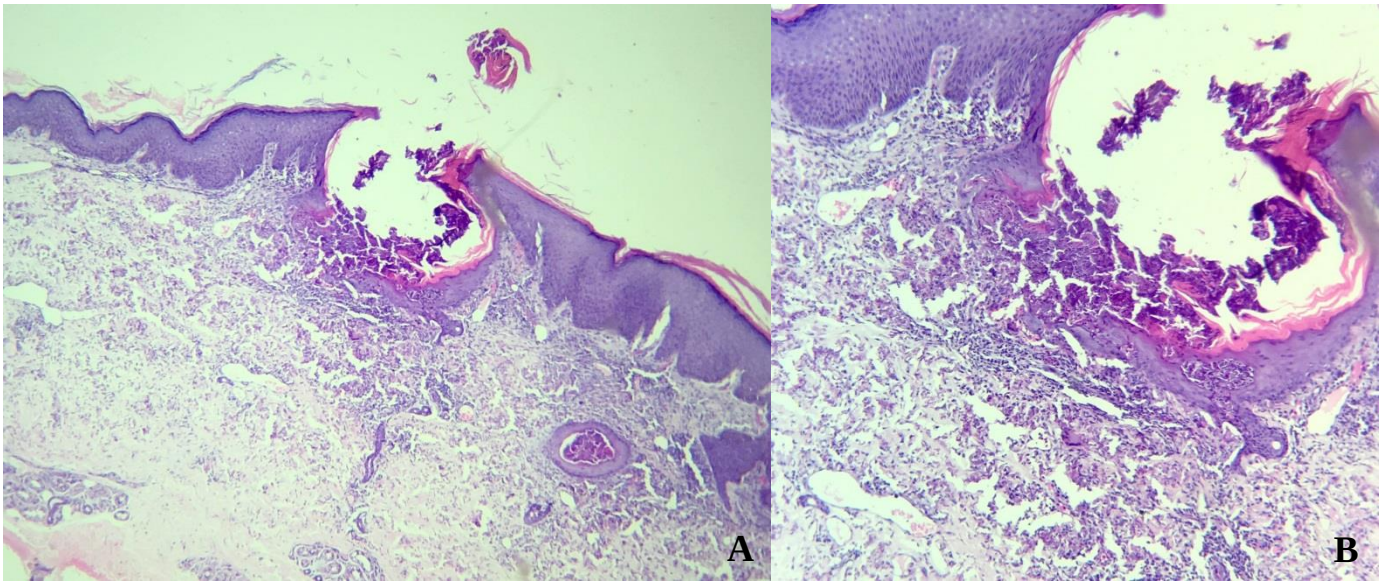

Figure 3. Abnormal, perforating elastic fibers. H&E, A, 40x, B 100x.

Staining with Verhoeff's Van Gieson stain confirmed curled and granular elastic fibers in the mid and lower dermis (Figure 4). Considering the clinical presentation (red brown papules and plaques with keratotic lesions in a periumbilical location in an obese multiparous woman with hypertension) the diagnosis of periumbilical perforating pseudoxathoma elasticum (PPPXE) was made. Histopathological findings confirmed the diagnosis.

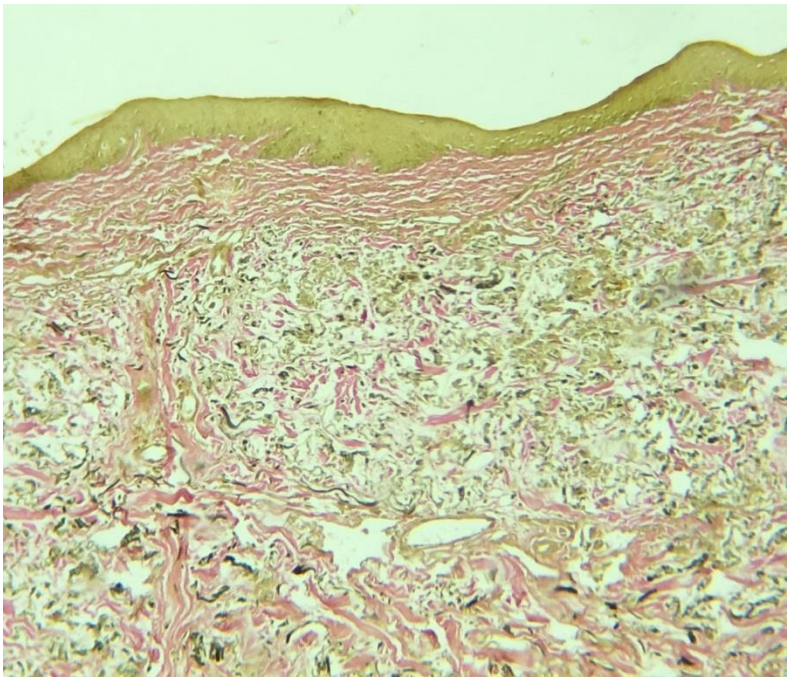

Figure 4. Verhoeff's Van Gieson stain confirmed the presence of abnormal elastic fibers, 100x.

Discussion

Periumbilical perforating pseudoxanthoma elasticum (PPPXE) is a non-inherited, localized skin disease characterized histologically by degeneration of elastic fibers of the mid dermis with extrusion of these calcified elastic fibers to the skin surface through a channel lined by acanthotic epidermis [1, 2]. It is classically seen in elderly, multiparous, obese, women [1, 2]. Initially, it was described as pseudoxanthoma elasticum (PXE) with coexisting elastosis perforans serpiginosa (EPS). Lund and Gilbert analyzed seven cases and established it as a separate entity. They found that elastic fibers in PXE are fragmented in the upper dermis only and remain unmineralized. On the other hand, elastic fibers of the mid and lower dermis are fragmented in PPPXE and become calcified [3]. Recently, the term "perforating calcific elastosis (PCE)" has been suggested to describe this condition in cases presenting in obese and multiparous women to emphasize the lack of systemic symptoms and avoid the ominous prognosis of PXE [4].

There are two schools of thought on the etiology of PPPXE. Some authors consider it to be a localized variant of PXE based on the presence of angioid streaks (22% cases) and distant flexural lesions with accompanying hypertension and diabetes [5, 6]. Others believe it to be an acquired dermatosis secondary to cutaneous trauma caused by obesity, multiple pregnancies, ascites, and surgery [1, 2].

PPPXE presents as a periumbilical plaque, of reticulated and atrophic yellowish surface, intermixed by an area of hyperchromia, which gives a wrinkled appearance to the lesion. The surface of the plaque is studded by keratotic papules. Unlike other perforating disorders (characterized by discrete umbilicated papules), PPPXE is typically characterized by clustered keratotic papules. Rarely, the lesions might affect areas other than the periumbilical region and involvement of the sides of the neck, upper arm, anterior chest, axillae, groin, back, and periareolar region have been reported [1, 2, 7]. Pruritus is a variable feature. Occasionally lesions may discharge "purulent" material. Systemic manifestations have been reported in association with PPPXE and include vascular disease (hypertension, claudication, diminished pulses, or angina), and ocular disease (angioid streaks) [7]. Histologically, it is characterized by short, gnarled, calcified, basophilic elastic fibers that are being transepidermally eliminated. The abnormal elastic fibers are noted in the mid and lower dermis. Chronic inflammatory cells are noted around areas of transepidermal elimination [1, 2, 8].

There is no established treatment for this rare disease. Sapadin et al. have reported a case of PPPXE associated with chronic renal failure and have noted complete clinical regression after 5 months of hemodialysis [5]. As the pathological process involved in PXE and PPPXE is similar, it is noteworthy to mention treatment response of PXE to dietary calcium restriction (800mg/day) and oral phosphate binder. Patients on dietary calcium restriction showed clinical regression in PXE lesions. Sherer et al. have tried oral phosphate binder (1800 mg of aluminum hydroxide tablets or liquid daily in 3 divided doses) in 6 patients and have reported significant clinical and histopathological improvement in 3 patients, usually beginning around month 4 of treatment [9]. The authors believe these treatment modalities may be tried in PPPXE, considering the response noted in PXE.

References

1. Muralidhar S. Periumbilical perforating pseudoxanthoma elasticum. Indian J Dermatol Venereol Leprol 1994;60:51-3.
2. Budania A, De D, Saikia UN, Kanwar AJ, Mahajan R, Yadav S. Periumbilical perforating pseudoxanthoma elasticum--an acquired perforating disorder. Int J Dermatol 2012;51(4):439-41. [PMID: 22435434]
3. Kocaturk E, Kavala M, Zindanci I, Koc M. Periumbilical perforating pseudoxanthoma elasticum. Indian J Dermatol Venereol Leprol 2009;75:329. [PMID: 19439905]
4. Lopes LC, Lobo L, Bajanca R. Perforating calcific elastosis. J Eur Acad Dermatol Venereol 2003 Mar;17(2):206-7. [PMID: 12705754]
5. Sapadin AN, Lebwohl MG, Teich SA, Phelps RG, DiCostanzo D, Cohen SR. Periumbilical pseudoxanthoma elasticum associated with chronic renal failure and angioid streaks--apparent regression with hemodialysis. J Am Acad Dermatol 1998;39:338-44. [PMID: 9703148]
6. Woo TY, Rasmussen JE. Disorders of transepidermal elimination. Part 1. Int J Dermatol 1985;24:267–9. [PMID: 3160676]
7. Bressan AL, Vasconcelos BN, Silva RS, Alves Mde F, Gripp AC. Periumbilical and periareolar perforating pseudoxanthoma elasticum. An Bras Dermatol 2010;85(5):705-7. [PMID: 21152799]
8. Hosen MJ, Lamoen A, De Paepe A, Vanakker OM. Histopathology of pseudoxanthoma elasticum and related disorders: histological hallmarks and diagnostic clues. Scientifica (Cairo) 2012;2012:598262. [PMID: 24278718]
9. Sherer DW, Singer G, Uribarri J, Phelps RG, Sapadin AN, Freund KB, Yanuzzi L, Fuchs W, Lebwohl M. Oral phosphate binders in the treatment of pseudoxanthoma elasticum. J Am Acad Dermatol 2005;53(4):610-5. [PMID: 16198780]