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Indian Journal of Postgraduate Dermatology

Case Report

Periumbilical Perforating Pseudoxanthoma Elasticum: A Case Report

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Received: 24 October 2024
Accepted: 05 May 2025
EPub Ahead of Print: 21 August 2025
Published: 19 September 2025

DOI
10.25259/IJPGD_229_2024

Quick Response Code:

ABSTRACT

Periumbilical perforating pseudoxanthoma elasticum (PXE) is a subtype of pseudoxanthoma elasticum (PXE). It is a rare and non-hereditary form of PXE. There is no difference in the clinical presentation of skin lesions (as well as histological appearance) of this localised acquired cutaneous PXE from the inherited variety. We report an interesting case of a 60-year-old female who presented with keratotic and pruritic plaques around the umbilicus without any systemic involvement. The histological features were consistent with periumbilical perforating PXE. Our case is unique owing to its rare occurrence.

Keywords: Case report, Perforating, Periumbilical, Pseudoxanthoma elasticum

INTRODUCTION

Periumbilical perforating pseudoxanthoma elasticum (PPPXE) usually presents with well-demarcated, hyperpigmented plaques with peripheral keratotic papules usually around the umbilicus. Pseudoxanthoma elasticum (PXE) is a multiorgan involvement disease with a spectrum varying from pure heritable form (involving elastic fibers of the integument [plucked chicken appearance of skin affecting lateral neck, followed by the cubital, knee pit, wrists, underarms, groin, and other flexural areas], eyes, and circulatory system with systemic involvement, and associated with mutations in the multidrug resistance associated protein 6, and early age of onset) to completely acquired localised form (without systemic involvement).[1] Few cases of the acquired form of PXE have been reported to have systemic involvement in the form of hypertension, diabetes, chronic kidney failure, renovascular hypertension and angioid streaks.[2,3] PPPXE patients with chronic kidney failure have been observed with an aberrant phosphate-calcium metabolism suggesting that cutaneous calcium deposition may lead to transepidermal elimination, as in tumour calcinosis. Furthermore, obesity and multiple surgeries may lead to elastic fibre degeneration in these patients. A typical patient of PPPXE is an overweight adult with Fitzpatrick skin type V, female with history of multiple pregnancies and surgery (abdomen) or trauma.[2] PPPXE is a connecting link between the hereditary and non-hereditary subtypes.

CASE REPORT

A 68-year-old, non-obese, multigravida female came to the outpatient department with complaints of itchy plaques around the umbilicus. She noticed flat papules (yellow) in the periumbilical region about 45 years ago. These papules gradually coalesced into plaques over time [Figure 1]. There was nothing significant in her medical and family history.

Figure 1: Periumbilical keratotic papules and nodules coalescing into plaques along with skin laxity.

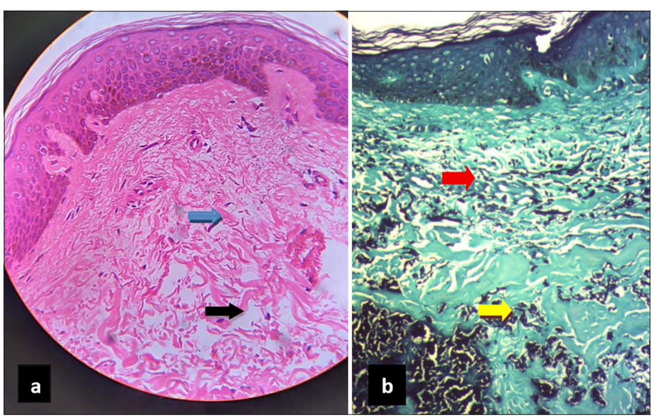

Figure 2: (a) Accumulation of the swollen and irregularly clumped elastin fibres was seen in the middle (blue arrow) and lower (black arrow) third of the dermis along with moderate perivascular lymphocytic infiltrate (haematoxylin and eosin, ×40). (b) Elastin fibres which stained deep black with Verhoeff–Van Gieson stain in the middle (red arrow) and lower third (yellow arrow) of the dermis were observed (×100).

On examination, a reticulated plaque of approximate size 15 cm × 13 cm with keratotic papules and nodules around the umbilicus, involving most of the abdomen, was noted. No similar lesions were seen on the neck, axillae and rest of the body. Routine laboratory tests were normal. Abdominal ultrasonography, echocardiography and fundus examinations were found to be normal. On histopathology examination, an accumulation of fragmented, clumped and curled fibres that stained deep black with Verhoeff–Van Gieson stain was seen in the lower and middle third of the dermis, along with moderate perivascular lymphocytic infiltrate [Figure 2a and b]. The patient was kept on emollients and antihistamines.

DISCUSSION

PPPXE and elastosis perforans serpiginosa (EPS) can be easily distinguished by histopathology, as in EPS, abnormal mineralised elastic fibres are present in the upper dermis, while in PPPXE, elastic fibres are fragmented, clumped and calcified.

We should differentiate the case of PPPXE from PXE-like papillary dermal elastolysis. The latter, on histopathological examination, shows decreased elastic tissue in a band-like distribution in the papillary dermis, unlike PPPXE.

Radical scavengers (antioxidants) might help in the treatment of this disorder as it has been observed in many reports that fibroblasts in this disease are affected by redox imbalance. So far, there is no established treatment for PPPXE. Oral phosphate binders and dietary calcium restriction (800 mg/day) have been associated with clinical improvement in PXE.[4] Oxidative stress has been shown to influence calcification, which is associated with PXE pathogenesis.[5] Involvement of other organs of the body and severity of complications decides the prognosis and life expectancy of this disease.

CONCLUSION

PPPXE is a subtype of hereditary PXE that is associated with transepidermal elimination. Further metabolic abnormalities, such as chronic kidney failure or parathyroid disorders, should be investigated in the absence of mechanical causes for perforating dermatosis, such as multiparity and surgeries.

Ethical approval: Institutional Review Board approval is not required.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship: Nil.

Conflicts of interest: There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation: The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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How to cite this article: Singh S, Das S. Periumbilical Perforating Pseudoxanthoma Elasticum: A Case Report. Indian J Postgrad Dermatol. 2025;3:163-5. doi: 10.25259/IJPGD_229_2024