Archives of Clinical & Experimental Dermatology

Yumed Text
OPEN ACCESS JOURNALS

Case Report | Vol 7 Iss 1
ISSN: 2583-6374
https://dx.doi.org/10.46527/2583-6374.182

Periumbilical Perforating Pseudoxanthoma Elasticum: A Rare Case Report

Shukla R, Ovhal AG, Chavanda KS and Darkase BA*

Department of Dermatology, Venereology & Leprosy Vilasrao Deshmukh Government Medical College Latur, India

***Corresponding author:** Darkase BA, MD, Department of Dermatology, Venereology & Leprosy, Vilasrao Deshmukh Government Medical College Latur, Maharashtra, Vikram Nagar Boudhi Chowk Latur, Maharashtra, India, Tel: +91-9405915340; E-mail: bhushandarkase@gmail.com

Received: April 28, 2025; **Accepted:** May 11, 2025; **Published:** May 20, 2025

Abstract

Periumbilical perforating pseudoxanthoma elasticum is a rare perforating dermatosis characterized by transepidermal elimination of abnormal elastic fibers with calcium deposition. It typically affects obese, middle-aged, multiparous women and presents as hyperpigmented plaques around the umbilicus. Although often limited to the skin, PPPXE may be associated with ocular and cardiovascular involvement. Fewer than 50 cases have been reported in the literature. We report a case of a 65-year-old multiparous woman with a 6-year history of pruritic abdominal lesions unresponsive to antifungals and corticosteroids. Clinical examination revealed irregular, atrophic scars with keratotic papules. Dermoscopy showed yellowish-brown structureless areas with central keratotic plugs. Histopathology demonstrated epidermal hyperplasia, hypergranulosis, parakeratotic columns, and transepidermal elimination of fragmented elastic fibers. Ophthalmologic evaluation revealed retinal pigment epithelium mottling. The patient had a history of hypertension but no family history of similar lesions. Etiology of PPPXE is not well established with some researchers considering it a localised cutaneous variant of pseudoxanthoma elasticum while others consider it a distinct clinical entity. This case highlights the rarity of PPPXE and underscores the importance of systemic evaluation, particularly ocular assessment, in suspected cases.

Keywords: ***Perforating; Periumbilical; Transepithelial; Pseudoxanthoma***

1. Abbreviations

PPPXE: Periumbilical perforating pseudoxanthoma elasticum; ECG: Electrocardiogram; PXE: Pseudoxanthoma elasticum; HIV: Human immunodeficiency virus; VDRL: Venereal Disease Research Laboratory

Citation: Shukla R, Ovhal AG, Chavanda KS, et al. Periumbilical Perforating Pseudoxanthoma Elasticum: A Rare Case Report. Arc Clin Exp Dermatol. 2025;7(1):182.

1. Introduction

Periumbilical perforating pseudoxanthoma elasticum (PPPXE) is a rare acquired skin disorder affecting the periumbilical area. The condition typically presents as a localised cutaneous dermatosis most often reported in obese, middle-aged, multiparous women. The disorder is characterised by the transepidermal elimination of altered dermal elastic fibres. PPPXE typically presents as gradually progressive, asymptomatic or mildly itchy erythematous or hyperpigmented papules in the periumbilical area, evolving into a central atrophic plaque with scaly margins and peripheral keratotic papules. Although typically confined to the skin, some patients have shown associated systemic conditions, particularly involving the cardiovascular system, The etiology of PPPXE is not well established with some researchers considering it a localised cutaneous variant of pseudoxanthoma elasticum while others consider it a distinct clinical entity, it is a rare disorder with less than 50 cases reported in literature [1]. There is currently no definitive treatment for PPPXE, topical steroids and retinoids have shown minimal to no benefit. Although therapies used in hereditary PXE, such as dietary calcium restriction and oral phosphate binders, have been considered due to shared pathology, their effectiveness in PPPXE remains unproven.

2. Case Report

A 65-year-old multiparous female presented with complaints of multiple itchy lesions on abdomen since 6 years. Initially she noticed a skin-colored soft papule on lower abdomen with subsequent development of multiple similar lesions over lower abdomen reaching towards lower margin of umbilicus. As new lesions developed, older lesions healed with formation of multiple atrophic scars of variable shapes. She has taken multiple courses of antifungals and topical corticosteroids from private clinics with no improvement. She was under treatment for hypertension. She did not give any history of difficulty in breathing or blood in vomitus, sputum, or stool. Her surgical and drug histories were also unremarkable. There was no history of similar lesions in her family. The patient showed no response to topical tretinoin and is planned to undergo CO_2 laser ablation. Her complete blood count, liver function tests, renal function tests, blood sugars, lipid profile, were within the normal limits. ECG, chest X-ray, and abdominal ultrasonography were normal. HIV, Hepatitis B, Hepatitis c, VDRL tests were negative.

Cutaneous examination revealed the presence of multiple ill-defined, irregular skin-colored atrophic scars of variable shapes coalescing to form a plaque on lower abdomen reaching towards lower margin of umbilicus with few keratotic papules at the margins (FIG. 1). The lesions were skin colored while few had a violaceous tinge, scarring was evident in the involved area. Dermoscopy revealed yellowish brown structureless areas along with semicircular, serpiginous/curved yellowish-brown lines, along with a keratotic plug with central crater (FIG. 2).

Histopathologic examination shows irregular epidermal hyperplasia with hypergranulosis, but at foci shows parakeratotic columns with underlying hypogranulosis, the dermis just below this foci shows increase in number of elastic fibres. The elastic fibres appear fragmented and crusted giving appearance of ravelled wool, at foci these altered elastic fibers appear perforating the epidermis (FIG. 3)

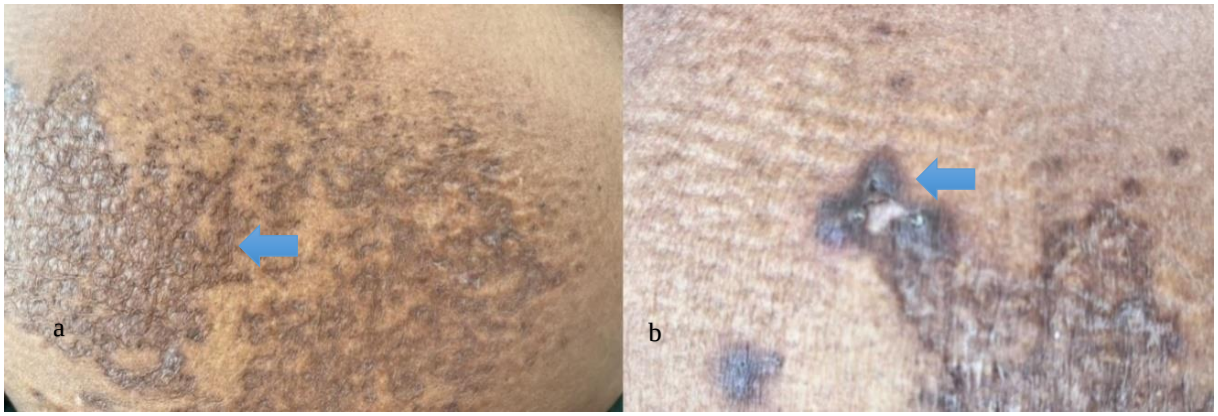

FIG. 1. **(a) Multiple ill-defined, irregular atrophic scars coalescing to form atrophic plaque on lower abdomen. (b) Keratotic papules at periphery of lesion.**

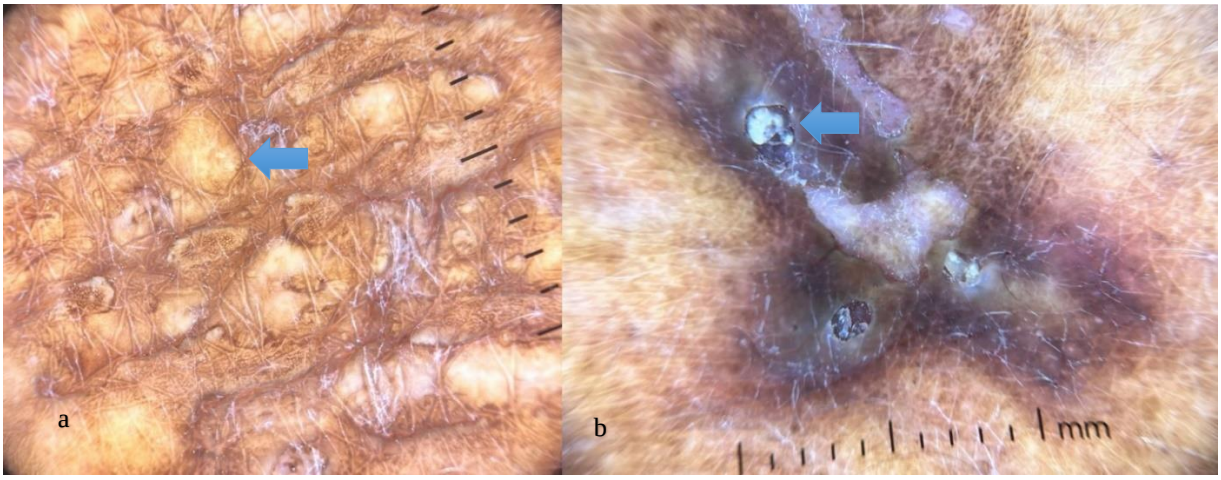

FIG. 2. (a) **Dermoscopy (10x) Yellowish brown structureless areas along with semicircular, serpiginous/curved yellowish-brown lines. (b) Dermoscopy (10x) Keratotic plug with central crater.**

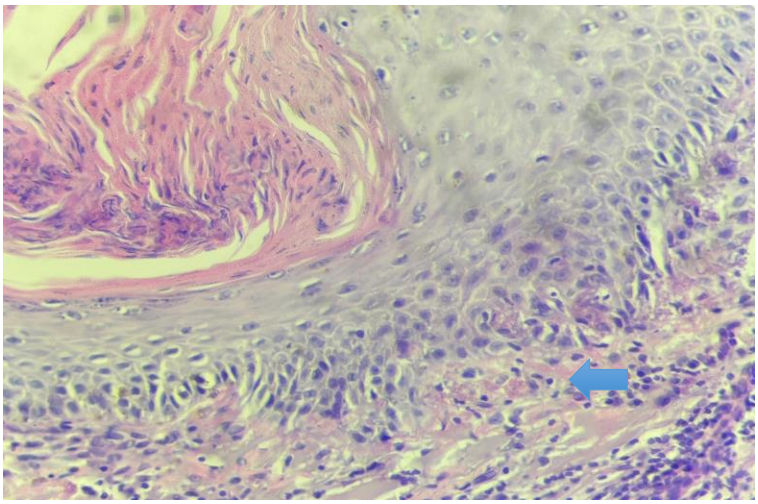

FIG. 3. **Elastic fibres appear fragmented and crusted giving appearance of ravelled wool (Hematoxylin & eosin stain: original magnification 40x).**

Ophthalmologic examination-Fundus image of right eye shows a normal cup disc ratio with peripapillary atrophy with choroidal tessellation. In temporal mid periphery of right eye, mottling of retinal pigment epithelium commonly known as Peau d'orange appearance is seen (FIG. 4).

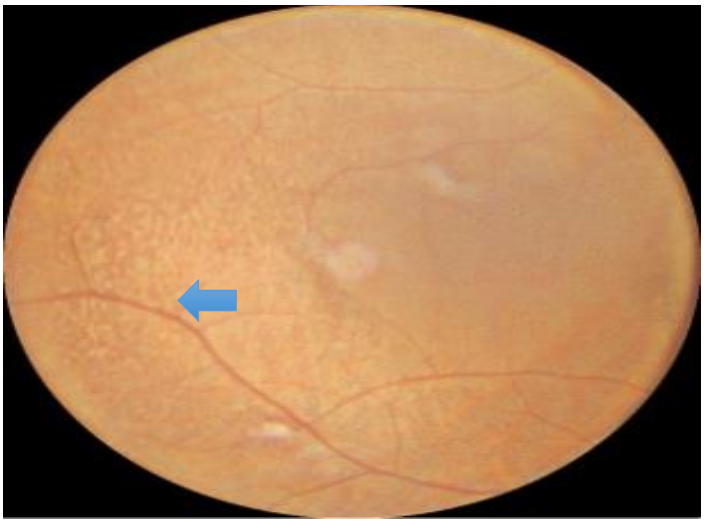

FIG. 4. **Fundus image of right eye, showing mottling of retinal pigment epithelium commonly known as Peau d'orange appearance.**

3. Discussion

Periumbilical perforating pseudoxanthoma elasticum (PPPXE) is a rare acquired skin disorder affecting the periumbilical area**,** it likely occurs due to repetitive mechanical stress on the abdominal wall, particularly in individuals who are obese, have undergone multiple pregnancies [2], persistent mechanical irritation, such as that caused by ascites, prior surgical interventions in the abdominal area, or repetitive trauma to the skin [3]. This chronic stretching may lead to degeneration and calcification of dermal elastic fibres. These altered fibres are subsequently eliminated through the epidermis via a process termed transepidermal elimination [2,4]. Histological analysis supports this, showing clusters of short, eosinophilic elastic fibres in the mid and upper dermis, often described as having a "ravelled wool" appearance, which are being extruded through the epidermis. These areas correspond to keratotic plugs that can be visualized through dermoscopy [5].

In one documented case, a patient with no systemic symptoms was found to have a heterozygous mutation in the ABCC6 gene, a mutation typically associated with hereditary pseudoxanthoma elasticum [6]. This observation raises the possibility that PPPXE may represent an intermediate form on the spectrum between acquired and inherited PXE. There is absence of ophthalmologic findings systemic involvement and hereditary features in most of the documented cases.Case reports published by Bum-Jin Juhn et al. described patient of PPPXE with cardiomegaly, atrial fibrillation [7], Robert allen Schwartz et al. [8] reported a case of PPPXE with asteroid hyalosis, angioid streaks and atrial fibrillation, few other case reports have described association with hypertension and angioid streaks [6] suggesting PPPXE to be a localised cutaneous manifestation of hereditary pseudoxanthoma elasticum. There is currently no definitive treatment for PPPXE, topical steroids and retinoids have shown

minimal to no benefit. Although therapies used in hereditary PXE, such as dietary calcium restriction and oral phosphate binders, have been considered due to shared pathology, their effectiveness in PPPXE remains unproven.

Pseudoxanthoma elasticum is an inherited condition that affects multiple organ systems and can lead to increased risk of morbidity and occasionally mortality. It is primarily marked by atypical mineral deposition primarily in skin, retina and cardiovascular system. Pseudoxanthoma elasticum is caused by mutations in the ABCC6 gene [9].

4. Conclusion

This case report describes typical cutaneous, dermoscopic and histopathological changes seen in periumbilical perforating pseudoxanthoma elasticum (PPPXE), along with eye involvement and chronic hypertension. These findings support the idea that PPPXE might be a localized skin form of the inherited condition pseudoxanthoma elasticum (PXE)Topical agents such as retinoids and steroids have shown limited success in treatment. The condition often persists, with lesions demonstrating slow progression or minimal response to therapy. We present a case of perforating periumbilical pseudoxanthoma elasticum accompanied by hypertension and ophthalmologic changes, aiming to highlight its rarity of presentation and workup required in suspected cases.

REFERENCES

1. Maronese CA, Spigariolo CB, Boggio FL, et al. Clinical, genetic, and ultrasonographic features of Periumbilical Perforating Pseudoxanthoma Elasticum. Skin Res Technol. 2021;27(4):646-7.
2. Kazakis AM, Parish WR. Periumbilical perforating pseudoxanthoma elasticum. J Am Acad Dermatol. 1988;19(2 Pt 2):384-8.
3. Lal NR, Bandhopadhyay D, Verma R, et al. Perforating calcific elastosis: Revisiting a rare entity. Indian J Dermatol 2018;63(2):186-8.
4. Mehta B, Amladi S, Nayak C, et al. An unusual periumbilical plaque. Indian J Dermatol Venereol Leprol. 2008;74(6):697-8.
5. Vishwanath T, Nagpal A, Shinde G. Periumbilical papules in a middle-aged woman. JAMA Dermatol. 2019;155(12): 1418-9.
6. Miller MK, Friedman RJ, Heilman ER. Degenerative diseases and perforating disorders. In: Elder DE, editor. Lever's Histopathology of the Skin. 11th ed. Philadelphia: Wolters Kluwer, USA; 2015. 458-78 p.
7. Juhn BJ, Sim WY, Lee MH. Periumbilical perforating pseudoxanthoma elasticum. Ann Dermatol. 1999;11(3):185-8.
8. Schwartz RA, Richfield DF. Pseudoxanthoma Elasticum With Transepidermal Elimination. Arc Dermatol. 1978;114(2):279-80.
9. ClinVar. https://www.ncbi.nlm.nih.gov/clinvar. Accessed on January 15, 2021