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PERIUMBILICAL PERFORATING PSEUDOXANTHOMA ELASTICUM: A RARE CASE REPORT WITH A DISTINCTIVE PRESENTATION

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ABSTRACT: Periumbilical perforating pseudoxanthoma elasticum (PPPXE) presents as well-defined, yellowish, atrophic plaques encircling keratotic papules, often observed in obese, multiparous women. This dermatosis is considered either a localized acquired cutaneous disorder or a variant of hereditary pseudoxanthoma elasticum. We report a case clinically diagnosed with differentials but confirmed as PPPXE through histopathological examination. The patient, a middle-aged woman, presented with characteristic lesions in the periumbilical region, prompting consideration of various differential diagnoses. However, microscopic analysis revealed the distinctive features of perforating pseudoxanthoma elasticum, including the presence of transepidermal elimination channels and abnormal elastic fibers. This case underscores the importance of histopathological evaluation in confirming the diagnosis of PPPXE, especially when clinical presentation overlaps with other cutaneous conditions. Awareness of this entity is crucial for accurate diagnosis and appropriate management, given its potential implications for systemic health. Further research is warranted to elucidate the pathogenesis and optimal treatment strategies for PPPXE. Our case contributes to the expanding literature on this rare dermatosis, emphasizing the significance of a multidisciplinary approach involving dermatologists and pathologists for its diagnosis and management.

INTRODUCTION: Perforating pseudoxanthoma elasticum (PPPXE) is a rare disorder. The localized lesion is prevalent predominantly in middle-aged, multiparous, obese black females [1-3]. It is a typical abdominal plaque that is situated over the umbilicus [1-4]. The plaque is a distinct, hyperpigmented lesion that has the potential to gradually grow in diameter [4]. Its surface has been characterized as being fissured and verrucoid or reticulated, grooved, & atrophic [3, 4].

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The edge of the plaque incorporates hyperkeratotic papules. A "purulent" secretion could be caused by the border of a plaque or papule being compressed. According to some investigators, the condition is an acquired dermatosis brought on by cutaneous injuries from previous pregnancies, adiposity, and many abdominal operations, as well as trauma that induced elastic fibre degeneration in the patient [1, 5].

PPPXE was viewed as a link between the pure inherited form and the pure acquired version [6]. Here, we report a case of a female clinically diagnosed with differential diagnosis as bowen's disease and lymphangioma circumscriptum over the periumbilical area but histopathologically proved as PPPXE which makes PPXE of a distinctive clinical presentation.

Case Report: A 48-year-old well built, multipara female presented with asymptomatic lesion which was dark colored and raised over the umbilical region since 8-9 months. Initially, appeared above the umbilical area which was yellowish to brownish in color of smaller size and progressed to the present size. Two months later, similar lesion appeared below the umbilical region, but the lesion was associated with multiple brownish-black color smaller sized lesions arranged in circular configuration. History of previous lower segment caesarean section (LSCS) present. There was no history of similar disease in the family. There was no history of similar lesions elsewhere over the body. Cardiologic evaluation (normal electrocardiogram and echocardiogram) and ophthalmoscopic examination did not show any changes. Routine blood investigations and serum lipid profile were all within normal range.

On Cutaneous Examination: Well defined hyperpigmented plaque with keratotic surface at centre with surrounding skin coloured patch present above the supraumbilical area with skin colored to brownish coloured plaque present over infraumbilical area with presence of annularly arranged hyperpigmented keratoticpapules, the largest plaque measuring approx 5*5cm^2 **Fig. 1.**

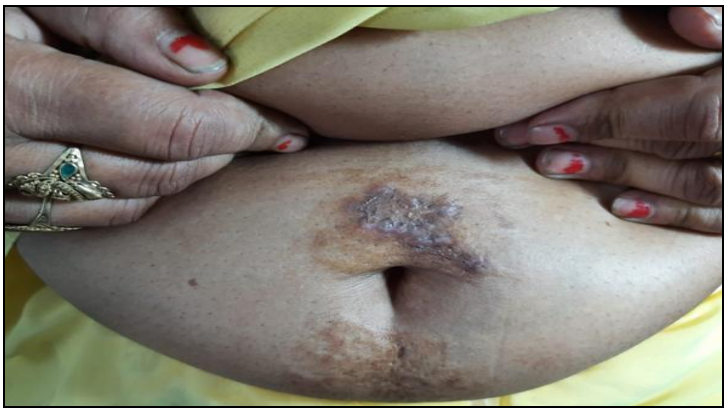

FIG. 1: WELL DEFINED HYPERPIGMENTED PLAQUE WITH KERATOTIC SURFACE AT CENTRE WITH SURROUNDING SKIN COLOURED PATCH PRESENT ABOVE THE SUPRAUMBILICAL AREA WITH SKIN COLOURED TO BROWNISH COLOURED PLAQUE PRESENT OVER INFRAUMBILICAL AREA WITH PRESENCE OF ANNULARLY ARRANGED HYPERPIGMENTED KERATOTICPAPULES, THE LARGEST PLAQUE MEASURING APPROX. 5*5CM^2.

Microscopic Examination: Revealed acanthosis, mild parakeratosis with mixed inflammatory infiltrate of eosinophils and lymphocytes. Foreign body giant cells were also seen. Focally epidermis is separated at dermo-epidermal junction. No epithelial dysplasia or granuloma seen **Fig. 2.** Special stain revealed black deposits of calcium on the altered elastic fibres which was consistent with the diagnosis of PPPXE **Fig. 3.**

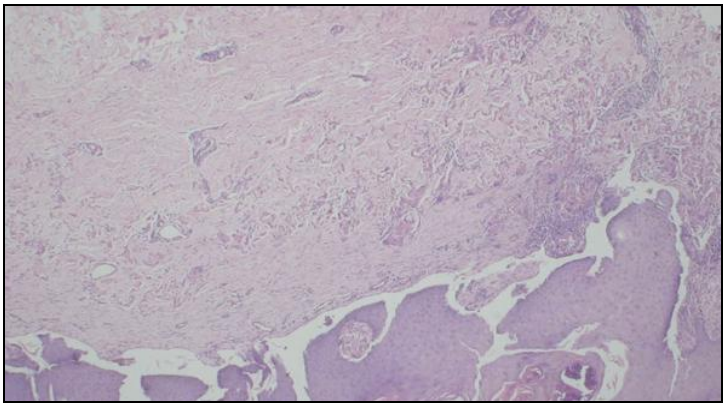

FIG. 2: ON HISTOPATHOLOGICAL EXAMINATION- ACANTHOSIS, MILD PARAKERATOSIS WITH MIXED INFLAMMATORY INFILTRATE OF EOSINOPHILS AND LYMPHOCYTES. FOREIGN BODY GIANT CELLS WERE ALSO SEEN. FOCALLY EPIDERMIS IS SEPARATED AT DERMO-EPIDERMAL JUNCTION. NO EPITHELIAL DYSPLASIA OR GRANULOMA SEEN

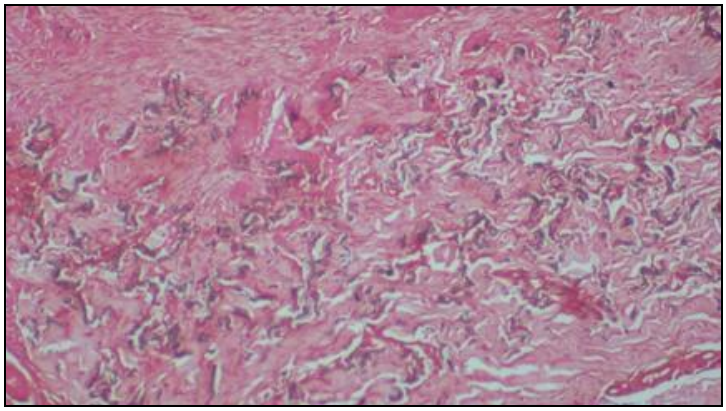

FIG. 3: SPECIAL STAIN REVEALED BLACK DEPOSITS OF CALCIUM ON THE ALTERED ELASTIC FIBRES

DISCUSSION: Earlier PPPXE was explained as pseudoxanthoma elasticum (PXE) with coexisting elastosis perforans serpiginosa (EPS). It was first founded as a separate entity by Lund and Gilbert [7]. PPPXE was also given the name as "localised acquired cutaneous pseudoxanthoma elasticum" since it was thought to be "acquired" and to lacking systemic involvement [8]. PPXE appears in areas of stress and movement. Early in the disease, the skin lines of the neck and axilla are accentuated. As the disease progresses, yellow papules appear in the flexural regions, such as the neck, axillae, inguinal folds, antecubital and popliteal fossae, and the periumbilical area. In our case, the patient was a black and multiparous women whose lesions were concentrated in the periumbilical region. Clinically, the usual sites of predilection were spared which was also seen in a study done by Maronese CA & hiscolleagues [9]. This makes our case report distinctive and conveys a message to all the budding dermatologist to make an opinion of suspecting PPXE especially in a black, multiparous female presenting with lesions over the periumbilical area with a varied presentation**.**

Lymphangioma circumscriptum presents on the skin surface as grapelike groups of thin walled, translucent, lymph-filled vesicles, often compared with frog spawn. Haemorrhage within the lesions can create a deep red or black appearance. In lymphangioma circumscriptum histopathological examination reveals acanthosis and hyperkeratosis of epidermis. Within the papillary and reticular dermis, there are dilated lymphatic channels containing esosinophilic proteinaceous material in the papillary dermis [10]. EPS may be idiopathic, induced by medications such as penicillamine or captopril, or associated with connective tissue diseases, including Marfan and Ehlers Danlos syndrome, cutis laxa, and pseudoxanthoma elasticum [10]. PPPXE may represent a localized variant of hereditary PXE, thereby rendering mandatory the same screening for the cardiovascular and ocular stigmata of the disease [11].

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CONFLICTS OF INTEREST: Nil

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