Underreporting of Perforating Osteoma Cutis: A Call for Recognition and Diagnostic Consistency

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Osteoma cutis is characterized by the presence of bone within the dermis or subcutaneous tissue. This disorder can occur in isolation or secondary to an underlying condition such as trauma, genetic syndromes, inflammatory processes, or neoplasia[1]. The exact incidence and prevalence of osteoma cutis is unknown, but it is thought to be uncommon, affecting less than 1% of the general population[2]. However, recent radiologic studies indicate cutaneous osteomas may be detected in as many as 42.1% of individuals[3].

As osteoma cutis is clinically considered a rare condition, its underlying pathophysiology remains unclear. Although several mechanisms have been posited, the most widely accepted theory involves the metaplastic transformation of fibroblasts into osteoblasts, possibly due to genetic abnormalities related to bone formation regulation[2]. Another proposed mechanism involves the differentiation of primitive mesenchymal cells into osteoblasts, which then migrate to an ectopic site[2].

Osteoma cutis can be classified based on its clinical presentation. Several variations have been reported and include solitary, widespread, plate-like, and multiple miliary[3]. Additionally, osteoma cutis is described as either perforating or non-perforating, with the former being considered the less prevalent form[5]. Diagnosis of osteoma cutis is typically confirmed through skin biopsy. Histopathological examination reveals dense eosinophilic deposits in the subcutaneous or dermal tissue, along with bony spicules that feature noticeable cement lines and calcifications[2]. In some cases, the spicules may perforate the epidermis via a process known as transepidermal elimination and appear as a central crater enclosed by squamous epithelium[4].

We read with interest the report by Zipperer et al. describing a case of perforating osteoma cutis. In the introduction, the authors emphasize the “exceptionally rare” nature of this dermatologic condition, citing only six previous reports[4]. We have also traditionally considered the perforating variant of osteoma cutis to be exceedingly unusual. However, in recent years, we have encountered this condition at such a frequency that we question whether this is truly the case.

The senior author of this letter recently reported a case of osteoma cutis penetrating through the epidermis, although he did not specifically categorize it as "perforating" osteoma cutis[6]. In addition, he has encountered multiple other cases of osteoma cutis perforating through the epidermis over the past several years. We present these novel cases observed between 2020 and 2022 herein. Thus, we suspect perforating osteoma cutis is underreported.

The first case is a 67-year-old woman with no significant past medical history who presented with a 0.5 cm crusted papule on the right distal calf (Figure 1A). The clinical differential diagnosis included invasive squamous cell carcinoma, irritated seborrheic keratosis, and basal cell carcinoma. Shave biopsy revealed centralized moderate compact serous, parakeratotic/ neutrophilic crust overlying epidermal erosion, and moderate irregular epidermal hyperplasia. The underlying papillary dermis contained compact pink bone with blue nuclei, characteristic of osteoma cutis. The bone appeared to be perforating through the epidermis and was surrounded by a mixed infiltrate of lymphocytes, neutrophils, and histiocytes (Figures 1B and 1C).

The second case describes a 73-year-old man with a past medical history significant for diabetes mellitus who presented with a 0.4 x 0.3 cm crusted papule on the left lateral posterior ankle (Figure 2A). The clinical differential diagnosis included squamous cell carcinoma, irritated seborrheic keratosis, basal cell carcinoma, and dermatofibroma. Shave biopsy revealed moderate to focally marked regular to irregular epidermal hyperplasia. The center of the specimen revealed moderate parakeratotic, serous, and neutrophilic crust overlying epidermal erosion. The papillary dermis in the center of the specimen contained a large focus of pink bone that was perforating through the overlying epidermis (Figures 2B and 2C).

Lastly, the third case is a 76-year-old woman with no significant past medical history who presented with a 0.3 cm crusted papule on the left elbow (Figure 3A). The clinical differential diagnosis included squamous cell carcinoma and irritated seborrheic keratosis. Shave biopsy revealed moderate irregular epidermal hyperplasia in the center of the specimen. The center of the specimen contained a large focus of moderate to marked parakeratotic, neutrophilic, and serous crust overlying pink bone extending from the superficial dermis to outside the epidermis (Figures 3B and 3C).

In all three instances, the diagnosis of perforating osteoma cutis was made. The presentation of these novel cases, along with the previously published case by the senior author, may suggest perforating osteoma cutis is underreported in the literature.

Potential reasons for this may be attributed to the benign nature of osteoma cutis and the clinical management of patients not being influenced by the distinction of perforating versus non-perforating subtypes[3]. Additionally, patients may fail to seek medical attention or intervention for the lesion if they do not consider it medically significant. Possible clinical and/or histopathological misdiagnosis of other conditions, such as ossified pilomatrixoma, keratoacanthoma, squamous cell carcinoma, etc., may also contribute[3]. Moreover, pathologists' lack of awareness or recognition of its rarity may be leading to its omission in literature. Further, pathologists may disregard the significance of the perforating nature and fail to include comments on its presence in their reports. Finally, it is possible that cases diagnosed as non-perforating osteoma cutis would actually show perforation through the epidermis if multiple deeper levels were performed, but pathologists may not order them due to the benign nature of the condition and unclear significance of epidermal perforation.

We encourage pathologists reading this letter to diagnose and report osteoma cutis with epidermal perforation as "perforating osteoma cutis" moving forward. In doing so, we can assess whether this variant is genuinely rare or merely underreported. Obtaining an accurate incidence would enable us to determine the significance of this phenomenon and explore possible associations with other conditions.

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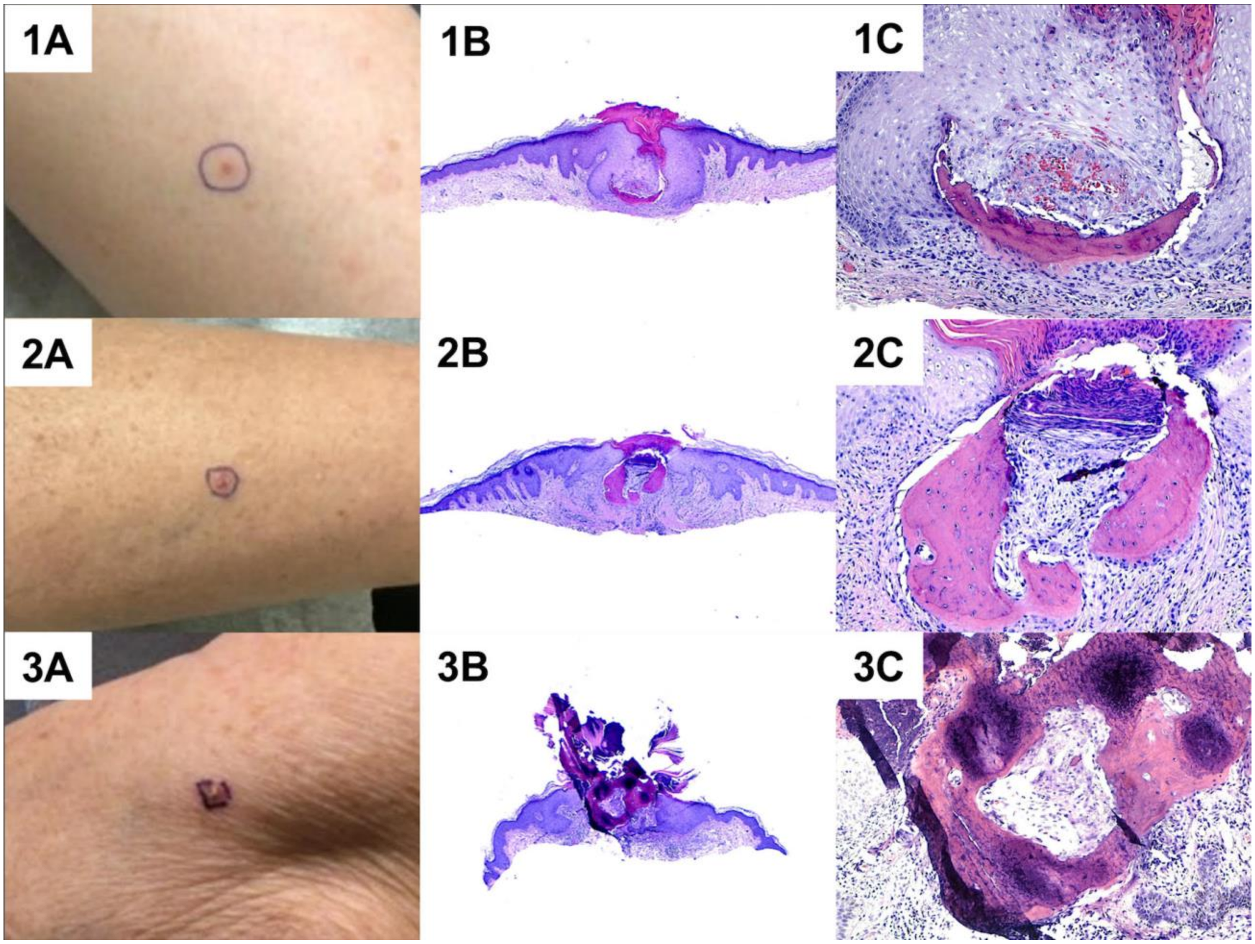

Figures 1-3. Clinical photos (A) and histology images (B, 20x, H&E; C, 100x, H&E) from three patient cases of perforating osteoma cutis as enumerated in the text.