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# Epidermolytic Hyperkeratosis and Congenital Platelike Osteoma Cutis in a Child

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*Epidermolytic hyperkeratosis is a rare congenital ichthyosis. Platelike osteoma cutis also is a rare diagnosis and is associated with abnormal ossification of cutaneous or subcutaneous tissue. A 17-month-old Hispanic girl presented with a plate of subcutaneous bone since birth as well as considerable scaling and hyperkeratosis centered around the joints. Histologic examination confirmed the diagnosis of both epidermolytic hyperkeratosis and osteoma cutis. Although there have been some cases of epidermolytic hyperkeratosis with other dermatologic conditions, we report a rare case of epidermolytic hyperkeratosis and platelike osteoma cutis.*

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**E**pidermolytic hyperkeratosis and platelike osteoma cutis are both uncommon diagnoses with characteristic clinical and histologic features. We report a case of a 17-month-old Hispanic girl who presented with epidermolytic hyperkeratosis and platelike osteoma cutis.

## Case Report

A 17-month-old Hispanic girl presented for evaluation of a rash that had been present since birth. The rash was unresponsive to topical steroids, as

prescribed by the patient's pediatrician. The parents reported that the patient was not particularly bothered by her rash and did not seem to scratch or pick at the lesions. The parents also noted a firm protuberance on the patient's left shoulder.

Physical examination revealed an infant in no apparent distress. Physical examination demonstrated thick hyperpigmented scaling in a bilateral distribution, most prominently located on the dorsum of the hands, dorsum of the feet, knees (Figure 1), elbows, and intermittently on her trunk. Evaluation of the left shoulder confirmed a 2×3-cm firm, well-defined, subcutaneous platelike plaque (Figure 2).

Histologic evaluation of the rash revealed hypergranulosis and prominent vacuolization of the superficial epidermis most consistent with epidermolytic hyperkeratosis (Figure 3). These findings of epidermolytic hyperkeratosis were seen in punch biopsies of the neck, upper extremity, and lower extremity. Histologic evaluation of the firm plaque on the patient's left shoulder showed foci of calcium deposition surrounded by thin portions of eosinophilic material and scattered osteoblasts, consistent with osteoma cutis (Figure 4).

Results of serum laboratory studies showed a parathyroid hormone level of 24.3 pg/mL (pediatric reference range, 10–65 pg/mL), an ionized calcium level of 4.7 mg/dL (pediatric reference range, 4.4–5.4 mg/dL), and an alkaline phosphatase level of 225 U/mL (pediatric reference range, 104–345 U/mL), all within reference range for an infant this age. The remainder of the patient's laboratory test results also were within reference range. Radiographic evaluation of the shoulder revealed subcutaneous calcification with no obvious connection to the underlying bony structures.

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The authors report no conflict of interest.

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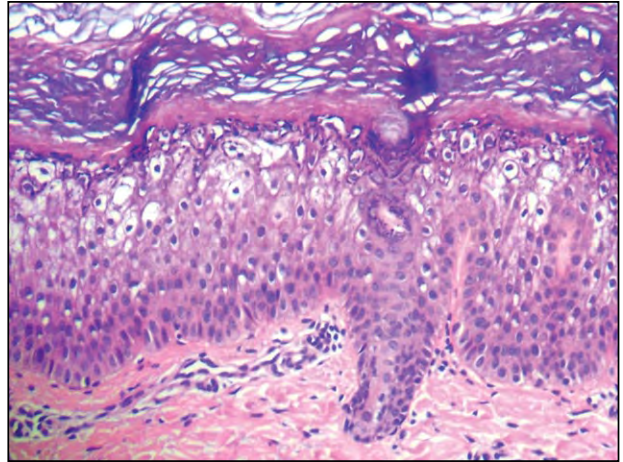
**Figure 1.** Mildly scaly hyperpigmented patches on the right knee with a subtle corrugated-appearing texture.



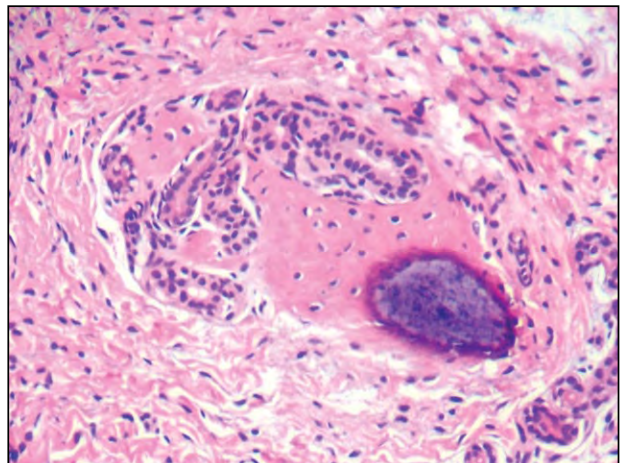
**Figure 2.** Outline of a firm subcutaneous platelike plaque on the left shoulder.

### Comment

Epidermolytic hyperkeratosis is caused by degeneration of keratinocytes due to a mutation involving keratin 1 and/or keratin 10,<sup>1</sup> which results in a disease characterized by hyperkeratosis, erosions, and peeling that diminishes the skin's ability to desquamate, retain water, and provide an adequate barrier to outside pathogens. It may occur either spontaneously or due to an autosomal-dominant mutation. The prevalence has been estimated between 1 in 200,000 and 1 in 300,000.<sup>2</sup> Treatment is aimed at decreasing potential breaks in the skin's role as a barrier to outside pathogens, which is achieved by avoiding epidermal trauma while using antimicrobial soaps and medications (topical or oral antibiotics) to decrease bacterial colonization. Some physicians target treatment at reducing the hyperkeratotic aspect of the disease by using keratolytic topical medications or increasing keratinocyte turnover by using vitamin A analogues (oral or topical retinoids) or vitamin D preparations.<sup>2</sup> However, one must be careful not to thin the epidermis with epidermolytic hyperkeratosis so aggressively that it impairs the skin's barrier function. The increased amount of transepidermal water loss must be treated with continual use of moisturizers and emollients.<sup>2</sup>



**Figure 3.** Punch biopsy demonstrating vacuolar degeneration in the stratum granulosum, consistent with epidermolytic hyperkeratosis (H&E, original magnification  $\times 100$ ).



**Figure 4.** Punch biopsy demonstrating foci of calcium deposition surrounded by portions of eosinophilic material and scattered osteoblasts, consistent with osteoma cutis (H&E, original magnification  $\times 100$ ).

Platelike osteoma cutis is a primary cutaneous ossification of dermal or subdermal tissue.<sup>3</sup> Unlike some other forms of cutaneous ossification, the origin or pathophysiologic mechanism of congenital platelike osteoma cutis is unknown at this time.<sup>4</sup> History of infection, trauma, neoplasm, or metabolic abnormality would rule out this idiopathic process. Treatment of osteoma cutis consists of surgical excision if the lesion is symptomatic or causes limitations in movement.<sup>4</sup>

It is possible that epidermolytic hyperkeratosis and congenital platelike osteoma cutis happened by chance in our patient. It is not inconceivable that an infant with underlying epidermolytic hyperkeratosis

had a mild amount of subclinical shoulder trauma or even a shoulder dystocia during delivery that resulted in a trauma-induced osteoma cutis.

However, we must consider the intriguing possibility of a unifying underlying pathogenesis. We focus our theory on the possibility of a localized abnormal calcium homeostasis. For many years, we have known that calcium plays an important role in regulating intercellular and intracellular processes, including gene transcription, translation, cell membrane channel regulation, and many other processes that ultimately result in normal keratinocyte differentiation. As keratinocytes normally progress through terminal differentiation, localized calcium levels are able to be maintained per the standard physiologic processes. However, as keratinocytes necrose in epidermolytic hyperkeratosis, an increase in localized calcium is created due to the intracellular calcium that is released into the extracellular environment, thereby disrupting the local physiologic calcium gradient. Whether or not this deregulation of calcium homeostasis in the epidermis translates to dysregulation of calcium homeostasis in the dermis is simply hypothetical.

Abnormal calcium and osseous metabolism has been proven to be involved in some patients with osteoma cutis, including patients with Albright hereditary osteodystrophy. Additionally, the discovery of the abnormal guanine nucleotide binding protein, alpha stimulating gene, *GNAS1*, has been noted in multiple ossifying disorders. This gene normally regulates osseous formation by inhibiting bone formation.<sup>5</sup>

The exact mechanism of extraskeletal bone deposition is still unknown. Some speculate that osteomas of the skin might arise from embryonic nests of pluripotent fibroblasts that can give rise to osteoblastic

cells.<sup>3</sup> Because osteoblasts are stimulated in physiologic bone formation by high serum calcium levels, one could speculate that localized high calcium levels may stimulate pluripotent fibroblasts to function as bone-forming osteoblasts.

For our patient, treatment has been targeted toward symptomatic management with continued use of moisturizers and emollients. She currently is being evaluated by the physical therapy department to determine the extent of her limitation caused by osteoma cutis. If it should be determined that her movement and future activities of daily living are or will become restricted, a surgical approach will be considered as a definitive treatment.

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