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Pathological Case of the Month

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THE PATIENT was a 9-month-old boy who had abnormal findings on an initial newborn screen for hypothyroidism, but a second test showed a normal circulating free thyroxine concentration, with serum thyroid-stimulating hormone concentrations that fluctuated between normal and slightly elevated. He was given replacement thyroid hormone to treat his compensated primary hypothyroidism. During the first few months of life, purplish, firm, nodular lesions were noticed on his abdomen and extremities (**Figure 1**), with the largest on the wrist and the knee (**Figure 2**). A biopsy specimen of one of the lesions on his abdomen had the microscopic appearance shown in **Figure 3**.

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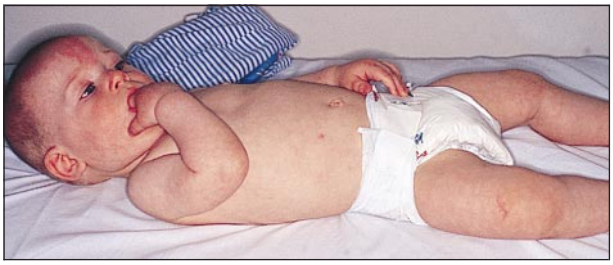

Figure 1.

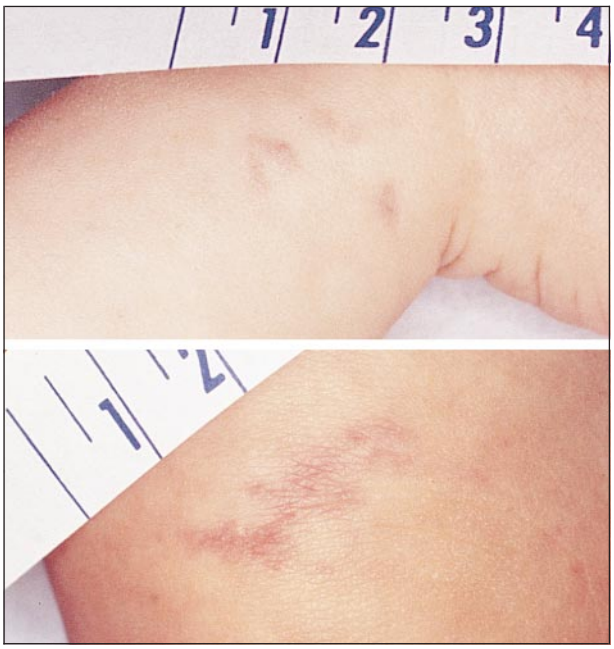

Figure 2.

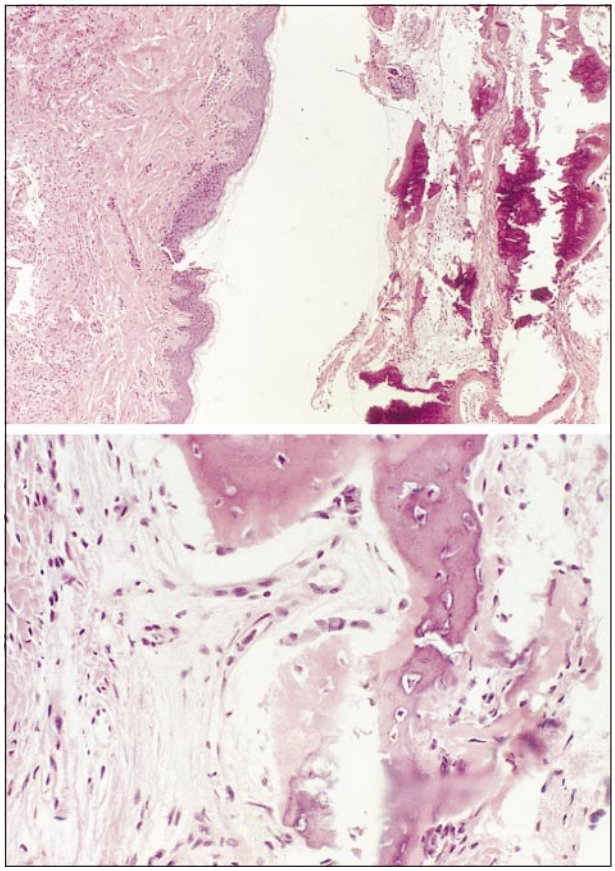

Figure 3.

Diagnosis and Discussion

Osteoma Cutis/ Pseudohypoparathyroidism

Figure 1. *Purplish, firm, nodular lesions were noticed on the patient's abdomen and extremities in the first few months of life.*

Figure 2. *The largest lesions appeared on the patient's wrist and knee.*

Figure 3. *Microscopic features from the biopsy specimen. Top, The epidermal, dermal, and subcutaneous tissues are shown, and the superficial epidermal and dermal tissues show no abnormalities (hematoxylin-eosin, original magnification ×40). Bottom, The features are shown at a higher magnification (×200), with the deep dermal and subcutaneous tissue to the right of a bony spicule containing osteocytes.*

The microscopic features from the biopsy are shown in Figure 3. Figure 3 (top) shows the epidermal, dermal, and subcutaneous tissues. The superficial epidermal and dermal tissues (top) showed no abnormalities. Within the deep dermal and subcutaneous tissues (bottom) there are spicules of bone formation. The features are also seen at higher magnification (×200) in Figure 3 (bottom), with the deep dermal and subcutaneous tissue to the right of a bony spicule that contains osteocytes. The bone is well mineralized. Cutaneous ossification can be a primary process or secondary process in which bone forms through metaplasia in a previous lesion.

Primary osteoma cutis is usually associated with pseudohypoparathyroidism (Albright hereditary osteodystrophy [AHO]), and in one series it was found in 42% of patients with AHO.[1] Purplish maculopapular lesions may be seen on the extremities, face, or trunk, with a firm, nodular consistency. Albright hereditary osteodystrophy should always be excluded when osteoma cutis is present, since it may be the presenting feature of this condition, especially in infancy. The metabolic features of AHO (decreased serum calcium and increased phosphorus and parathyroid hormone concentrations) reflect peripheral resistance to parathyroid hormone and may not be present in infancy. Osteoma cutis may also precede the characteristic phenotype (short stature, short 4th and 5th metacarpals, round face) by years.[2]

Since AHO is usually due to a defect in the Gs protein associated with cell surface receptors for polypeptide hormones, the patient may have resistance to more than 1 hormone. This may include resistance to serum thyroid-stimulating hormone that results in primary, overt,[3,4] or compensated hypothyroidism,[5] as was seen in our patient. Other children with AHO may have resistance to growth hormone–releasing hormone, with subsequent growth hormone deficiency,[6] and/or resistance to gonadotropins, resulting in gonadal insufficiency.[7,8]

Our patient lacked any of the phenotypic features of AHO and had normal motor and language development for his age. On further testing, he was found to have normal serum concentrations of calcium, phosphorus, and alkaline phosphatase, but a considerably elevated serum parathyroid hormone concentration. This was compatible with a state of peripheral resistance to parathyroid hormone (pseudohypoparathyroidism) that was compensated and confirmed the diagnosis of AHO. The presence of both compensated resistance to serum thyroid-stimulating hormone and parathyroid hormone in our patient is similar to a case in an older child with AHO reported by Coutant et al.[5]

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