

# Exaggerated Facial Lines on the Forehead and Cheeks

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A 36-year-old man presented to the emergency department with an olecranon fracture after falling from a tree. The patient had a medical history of type 2 diabetes mellitus and a surgical history of facial cosmetic surgery. He underwent internal fixation with orthopedic surgery for the olecranon fracture, and dermatology subsequently was consulted due to diffuse skin changes on the face. He reported that these dermatologic changes began around 17 years of age and had progressed to the current presentation. He denied itching, burning, pain, or contact with aradillos. A family history revealed the patient's brother also had a similar appearance. Physical examination revealed exaggerated facial lines on the forehead (top) and cheeks. Digital clubbing and skin thickening were noted on the hands (bottom) and feet; examination of the back revealed multiple hypopigmented patches. Observation of the scalp showed multiple symmetric ridges and grooves with sparse overlying hair consistent with cutis verticis gyrata. A punch biopsy of the forehead was obtained as well as bone radiography taken previously by the primary team.

## WHAT'S YOUR DIAGNOSIS?

- acromegaly
- cutaneous T-cell lymphoma
- lepomatous leprosy
- pachydermoperiostosis
- scleromyxedema

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The authors report no conflict of interest.

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doi:10.12788/cutis.0598

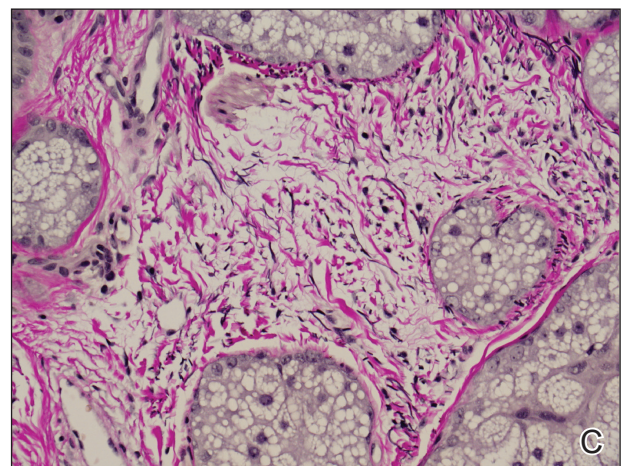
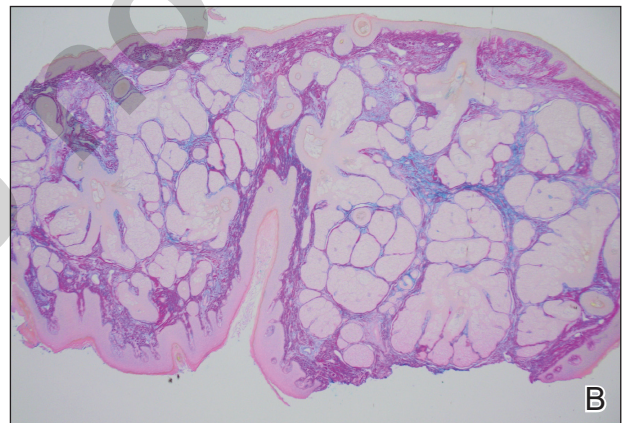
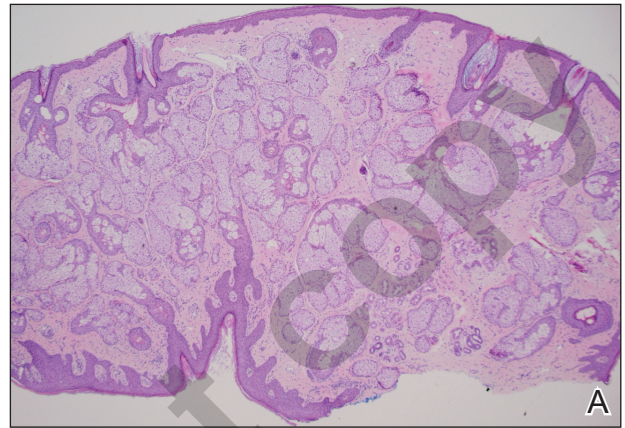
## THE DIAGNOSIS: Pachydermoperiostosis

**H**istopathology of the forehead punch biopsy demonstrated sebaceous hyperplasia with an occupation rate of greater than 40%, increased mucin, elastic fiber degeneration, and fibrosis. Pachydermia is graded from 0 to 3 depending on the degree of these changes; our patient met criteria for grade 3 pachydermia (Figure 1). Radiography revealed diffuse cortical thickening of the long bones that was most marked in the left femur (Figure 2); however, no other findings were demonstrative of Paget disease.

Pachydermoperiostosis (PDP) (also known as Touraine-Solente-Golé syndrome or primary hypertrophic osteoarthropathy) is a rare genetic condition that affects both the dermatologic and skeletal systems. Clinical features of the disease include progressive thickening and furrowing of the skin on the scalp and face (known as pachydermia), digital clubbing, and periostosis. Other potential cutaneous features include seborrhea, acne, hyperhidrosis of the palms and soles, cutis verticis gyrata, eczema, and a burning sensation of the hands and feet. Myelofibrosis and gastrointestinal abnormalities also have been reported.<sup>1</sup>

The disease typically affects males (7:1 ratio); also, men typically display a more severe phenotype of the disease.<sup>2</sup> It most commonly begins during puberty and follows a generally progressive course of 5 to 20 years before eventually stabilizing. Both autosomal-dominant with incomplete penetrance and recessive inheritance versions of PDP can occur. Prostaglandin E<sub>2</sub> (PGE<sub>2</sub>) has been implicated in the pathogenesis of PDP; PGE<sub>2</sub> is important in the inflammatory response and may evolve from disrupted protein degradation pathways.<sup>3</sup> Sasaki et al<sup>4</sup> additionally reported that the severity of pachydermia clinically and histologically appeared to correlate with the serum PGE<sub>2</sub> levels in affected patients. Prostaglandin E<sub>2</sub> causes a vasodilatory effect, perhaps explaining the clubbing observed in PDP, and also modifies the activity of osteoblasts and osteoclasts, causing the bone remodeling observed in the disease.<sup>4</sup>

In our patient, the initial differential diagnosis included PDP, as well as lepromatous leprosy, acromegaly, Paget disease of the bone, amyloidosis, scleromyxedema, and cutaneous T-cell lymphoma. However, the time course of the disease, lack of numerous symmetric thickened plaques and madarosis, and pathology argued against lepromatous leprosy. Acromegaly was ruled out due to lack of macroglossia as well as laboratory analysis within reference range including IGF-1 levels and thyroid function tests. Biopsy findings ultimately ruled out amyloidosis and cutaneous T-cell lymphoma. The bone scan revealed diffuse cortical thickening consistent with PDP,



**FIGURE 1.** A, Histopathology of a forehead biopsy showed increased sebaceous gland occupation (H&E, original magnification  $\times 4$ ). B, Colloidal iron stain demonstrated increased mucin (original magnification  $\times 4$ ). C, Verhoeff-van Gieson stain showed elastic fiber degeneration (original magnification  $\times 40$ ).





**FIGURE 2.** Radiography of the left femur demonstrated diffuse cortical thickening.

and there were no other radiologic findings suggestive of Paget disease.

Pachydermoperiostosis is diagnosed using the Borochowitz criteria, which entails that 2 of the following 4 fulfillment criteria must be met: familial transmission, pachydermia, digital clubbing, and/or bony involvement with evidence of radiologic alterations or pain. Our

patient met all 4 criteria. The clinical manifestations of PDP are variable with respect to skin and bone changes. The various clinical expressions include the complete form (ie, pachydermia, cutis verticis gyrata, periostosis), the incomplete form (ie, absence of cutis verticis gyrata), and forme fruste (ie, pachydermia with minimal or absent periostosis).<sup>5</sup>

Management for PDP involves surgical correction for cosmesis as well as for functional concerns if present. Symptoms secondary to periostosis should be managed with symptomatic treatment such as nonsteroidal anti-inflammatory drugs. Patients managed with etoricoxib, a COX-2-selective nonsteroidal anti-inflammatory drug, have had normalized inflammatory markers that resulted in the lessening of forehead skin folds. Oral aescin has been shown to relieve joint pain due to its anti-edematous effect.<sup>6</sup> Our patient received treatment with nonsteroidal anti-inflammatory drugs for symptomatic management of the associated joint pain but unfortunately was lost to follow-up.

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