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CASES FOR DIAGNOSIS

Skin and Bone Lesions in an Adolescent

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Clinical History

A 13-year-old boy was seen for a history of asymptomatic skin lesions on the trunk and legs; the lesions had increased in number progressively since 6 months of age. The mother denied the presence of similar lesions in other members of the family.

Physical Examination

A number of yellowish plaques were observed on the abdomen, in the lumbar region, and on the thighs (Figure 1). The lesions were slightly elevated and had an elastic consistency.

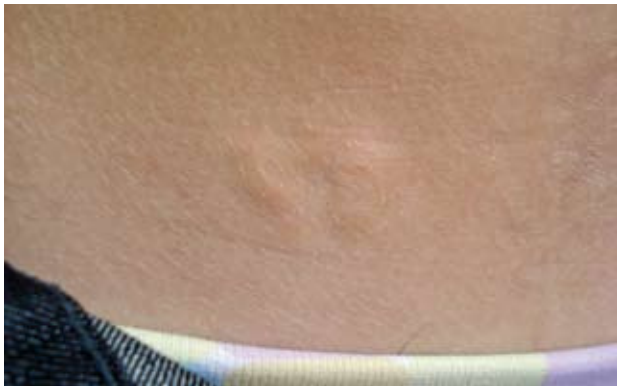

Figure 1.

Additional Tests

The only finding in the complete blood count and serum biochemistry was an alkaline phosphatase value of 322 U/L (a normal value for the age of the patient). The radiologic study (bone scan) performed at 4 years of age showed no abnormalities; however, a repeat scan at 13 years showed multiple, small, round foci of bone sclerosis, situated symmetrically in the carpal bones (Figure 2), the tarsi, and the pelvis.

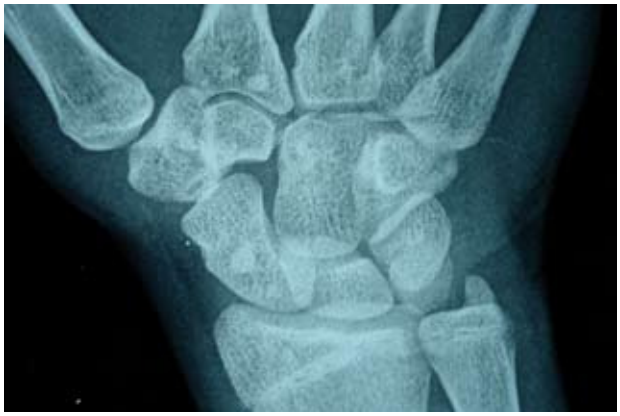

Figure 2.

Histopathology

Biopsy of one of the lesions showed a skin of normal appearance with hematoxylin-eosin stain. However, orcein staining revealed a normal papillary dermis and a marked increase in elastic fibers affecting the full thickness of the middle and deep dermis; many of these fibers were abnormally thick (Figure 3).

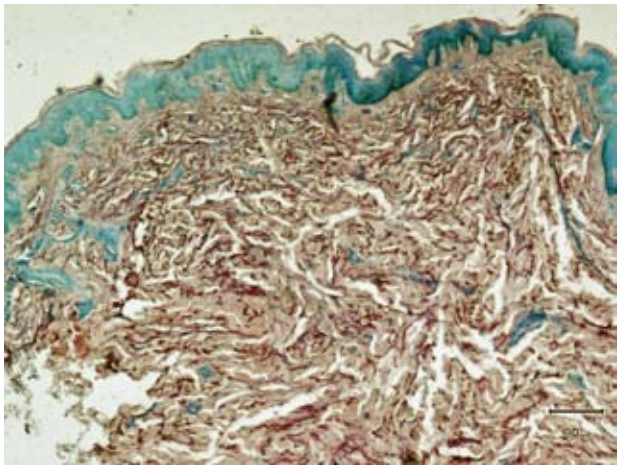

Figure 3. Orcein stain, ×40.

What Was the Diagnosis?

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Diagnosis

Buschke-Ollendorff syndrome.

Clinical Course and Treatment

At the time of writing, the patient had a normal growth and bone development, associated with a progressive appearance of new skin lesions but no new radiologic lesions.

Discussion

Buschke-Ollendorff syndrome is a rare condition characterized by the association of skin lesions called dermatofibrosis lenticularis disseminata, and osteopoikilotic bone lesions.[1] Its etiology is poorly understood, although recent research suggests that the underlying defect is due to heterozygosity for mutations that cause a loss of function of the *LEMD3* gene, which codes for an internal nuclear membrane protein.[2] It has autosomal dominant inheritance with variable expression and incomplete penetrance, although cases with no family history have been reported,[1,3,4] and also incomplete forms with skin lesions but with no associated bone lesions or vice versa.[3]

Dermatofibrosis lenticularis disseminata presents clinically as yellowish papules or plaques that are usually asymptomatic; they tend to appear during childhood, although cases occurring in adult life have also been reported.[3] The most common sites are the lumbar region, buttocks, arms, thighs, and abdomen, and the distribution may be symmetric or asymmetric.[3] From a histopathologic point of view, the lesions consist of connective tissue nevi; several types of these lesions have been described in patients with this syndrome, including elastic nevus, collagenomas, and lesions in which abnormalities of collagen and of elastic fibers coexist.[5]

Osteopoikilosis (spotted bones) is a polyostotic, polytopic osteosclerotic dysplasia that is completely asymptomatic, meaning that it is usually a incidental finding. It is characterized radiologically by the presence of well-delimited, round foci of bone sclerosis less than 10 mm in diameter, not affecting the cortical bone; they have a symmetrical distribution and are preferentially situated on the epiphyses and metaphyses of tubular bones, the scapulae, and the carpal, tarsal, and pelvic bones. Histologically there is an increase in the number and thickness of the trabeculae of the spongy bone.[4] Three types of osteopoikilosis have been described (punctate, striate, and mixed), the most common of which is punctate.[4] However, osteopoikilosis is not found exclusively in Buschke-Ollendorff syndrome; it has also been reported in association with other processes such as fibromas of the oral mucosa, keloids, histiocytosis, certain palmoplantar keratodermas, atrophic striae, morphea-like lesions, and systemic sclerosis.[3-5]

The laboratory findings are nonspecific, although some authors have reported disturbances of calcium and phosphate metabolism in patients with osteopoikilosis and in their first-degree relatives, including elevated alkaline phosphatase, parathyroid hormone, or 25-hydroxyvitamin D.[6]

Buschke-Ollendorff syndrome has been associated with other abnormalities, such as growth delay, supernumerary ribs or vertebrae, mental retardation, neuropathy, diabetes, or rheumatoid arthritis.[1,4] However, the prognosis of the disease is generally benign and specific treatment is not required.

Conflicts of Interest
The authors declare no conflicts of interest.

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