PAEDIATRIC DERMATOLOGY

BUSCHKE-OLLENDORFF SYNDROME: DIFFICULTIES IN DIAGNOSIS.

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Background: Buschke-Ollendorff syndrome (BOS) is a rare congenital autosomal dominant hereditary condition manifested by sclerotic, osseous formations seen on X-ray called osteopoikilosis and fibrous skin papules either elastin-rich (elastoma) or collagen-rich (dermatofibrosis lenticularis disseminata).
Skin manifestations usually occur combined with changes in bones, whereas some patients develop either skin or bone symptoms.
We present a 6-year-old male patient who was hospitalised in the Deprtment of Dermatology, Pediatric Dermatology and Oncology, Medical University of Lodz, Poland with connective tissue nevi localized in the sacrolumbar region and on the lower extremities. The nevi were slightly elevated and flattened, yellowish and painless. At the age of 2 he was firstly diagnosed as localized scleroderma and since histopathological examination excluded this condition another skin biospy was obtained. Second biospy revealed deposits of histiocytes among the muscle fibers and suggests occurance of storage disease. Based on results of additional tests and clinical manifestation histiocytosis was also excluded. Meanwhile patient started to suffer from strong nocturnal pain localized in both lower extremities. An X-ray showed spots of increased bone density and sclerotic cartilages,while genetic testing revealed a heterozygous loss-of-function mutation in the LEMD3 gene.

Key Message: Although Buschke-Ollendorff syndrome generally follows a benign course, it is important to avoid misdiagnosis and unnecessary treatment. Awareness of the possible concominant disorders such as otosclerosis with hearing impairment, stenosis of the aorta or diabetes should result in long-term and profound observation of patient with diagnosis of BOS.