

## An Unusual Presentation of Hereditary Multiple Exostoses

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### ABSTRACT

Hereditary multiple exostoses (HME) is an autosomal dominantly inherited disease characterized by multiple benign endochondral bone formations in the metaphysis of bones. Although most patients are asymptomatic, exostoses might cause pain and result in difficulty in movement, orthopedic deformities, growth retardation, short stature and chondrosarcomas. Herein, we report an 11 year old boy who applied to our dermatology outpatient clinic with swellings on the proximal nail folds of two fingers and finally diagnosed with HME.

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**Key words:** Hereditary multiple exostoses; Nail finding; Malignancy

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### LETTER TO THE EDITOR

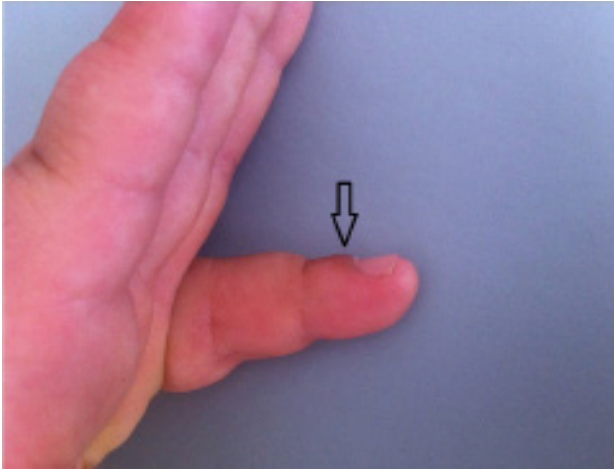
An 11-year-old male presented with nail deformity, swelling of the proximal nail folds on the index finger of the right hand and the third finger of the left hand. The lesions have developed slowly in the last two years and they were painless. Physical examination showed nail plate dystrophy, longitudinal ridges on the nails and firm, nontender nodules on the nail folds (Figure 1). A nodule, approximately 7 cm in diameter, was palpated on the proximal lateral side of the right lower leg. The child was healthy in general and the laboratory studies revealed normal findings. Relatedly, the patient's father suffered from the same skeletal abnormalities and passed through several surgical procedures.

X-ray examination revealed multiple exostosis on the distal phalanges of the affected fingers and on the right fibula and on both femur (Figure 2). The diagnosis of hereditary multiple exostoses (HME) was made clinically and radiologically. The patient was referred to the department of orthopaedic surgery where they confirmed the diagnosis. Symptomatic patients require surgical intervention. Our patient was asymptomatic and thus no surgery was performed.

HME is an autosomal dominantly inherited disease characterized by multiple benign endochondral bone formations in the metaphysis of long bones. Most patients are asymptomatic but exostoses might cause pain, difficulty in movement, orthopedic deformities, growth retardation and short stature. In addition, chondrosarcomas may develop in 1% to 25% of such cases<sup>[1,2]</sup>.

Few cases of HME presented with nail deformities were reported in the literature<sup>[1,3-5]</sup>.

Del-Rio *et al* reported a 14 year old boy presented with nail deformity and swelling of the nail fold on several fingers. X-ray examination revealed multiple exostoses on distal phalanges and also in long bones. His father and other relatives have suffered from the same disease but had no nail findings<sup>[1]</sup>.



**Figure 1** Palpable nodule on proximal nail fold of the finger (arrow).



**Figure 2** a: Exostoses on distal phalanges of the third finger of the left hand (arrow); b: Exostoses on the leg bones (arrows)

Yanagi *et al* reported a 16 year old female with a nail deformity and swelling in the right index finger. She has been suffering from HME and several operations were performed on her since childhood.

An exostosis of the distal phalanges was found by X-ray examination and the exostosis was removed surgically<sup>[3]</sup>.

A 9 year old boy with the swelling of several nails and proximal nail folds was reported. X-ray examination demonstrated multiple exostoses of the phalanges and of the other bones<sup>[5]</sup>.

Herein, we report an unusual presentation of HME with nail deformity. We believe that, the recognition of the disease by dermatologists is important because nail findings may be the only physical manifestation of HME.

## CONFLICT OF INTERESTS

There are no conflicts of interest with regard to the present study.

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