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A Case With Multiple Hereditary Exostosis

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Figure 1. Appearance of bony "lumps" on left iliac bone, on both forearms, on the legs and ulnar deviation of forth finger on the left hand.

Multiple hereditary exostosis is a relatively uncommon autosomal dominant disorder in which the clinical

hallmark is the growth of bony protuberances from particularly long bones resulting in a variety of orthopedic deformities and that of short stature. De novo, as in our patient whose first degree relatives showed no abnormality in screening, it also occurs with an estimated rate of $6.3-9.1 \times 10^{-6}$ (1). There is a definite excess of males within the entire affected population. This may partly be due to males having more severe and frequent complications. Herein, we present here a 15-year-old boy with multiple hereditary exostosis including scintigraphic findings.

A 15-year-old boy referred with complaints of several, solid masses in the bones, deformities in the extremities and short stature (Fig 1). The parents are nonconsanguineous. The first protuberance had appeared in left iliac bone at the age of two, followed by the upper and lower extremities and ribs. Another finding was ptosis on the left eye beginning from preschool years. Physical examination revealed multiple hard "bumps" on several bones including left hemipelvis, bilateral distal radius, ulna, both scapula, right proximal humerus, bilateral proximal fibula and that of distal tibia. His height, weight values were 144 cm and 35 kg respectively.

Deformities in the region of forearm and leg were also noted. X-ray survey showed a number of exostosis in the following bones; both scapula, right and left proximal humerus, right distal radius, right scapula, left second and forth finger metacarp, left fifth rib, right iliac anterior region, bilateral distal and proximal femur, bilateral proximal fibula, bilateral distal tibia and right proximal tibia. Bowing of ulna on the left, and subluxation of the hip on the right and deformation of femur were detected

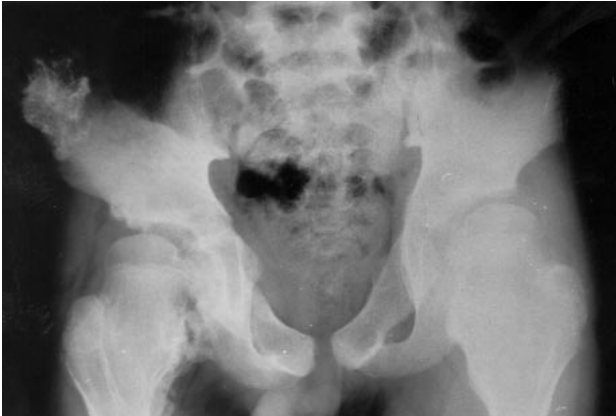


Figure 2. Pelvic radiograph showing marked exostosis in the right hemipelvis.

(Fig 2).

Scintigraphy with Technetium-99 m revealed markedly increased uptake in the pelvis iliac anterior region and to lesser extent in the areas of exostosis.

Hereditary multiple exostosis is a hereditary disease with an autosomal dominant trait. Since first degree relatives showed no abnormality in screening, de novo mutation is, of course, the cause of the disease in our case.

The disease is frequently accompanied by disturbances such as abnormal tabulation of bones, producing broad

and blunt metaphyses, and sometimes bowing of the long bones and shortening of ulna producing ulnar deviation of the hand. Furthermore, lower extremity deformities are also found. In our patient proximal exostosis of femur caused acetabular deformity resulting in subluxation of the right hip (2, 3). Hand involvement is relatively uncommon in hereditary multiple exostosis; in this patient metacarpals in left hand and the right scaphoid showed exostosis (4). Particularly, scaphoid involvement is found to be extremely rare. However, metacarpal lesions in hands do not usually cause functional deformity. Although malignant transformation, frequently into low grade chondrosarcoma, can occur in approximately 1-2% of affected young patients, no such transformation has been reported in adults. Pain and enlarging mass were considered as the major symptoms for malignancy. A single radiograph and scintigraph are insufficient to reveal malignancy. A sudden marked increase in the uptake of isotope with sudden onset of growth in a previously quiescent exostosis indicate possible existence of malign process. Therefore, the patients should be monitored with serial scintigraphic and tomographic investigations for the early detection of malignancy as we planned to follow our case (5). Bone scintigraphy in the evaluation of exostosis has been shown to be useful in assessing the level of active ossification and in detecting new areas. Since there is no specific treatment for the disease, profound deformities and progressive malign tumors can be treated by surgical intervention.

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