

Clinical Image

Hereditary Multiple Exostoses: Metaphyseal Location of Outgrowths in Two Siblings (Knees) – Distinction from Spondyloepiphyseal Dysplasia

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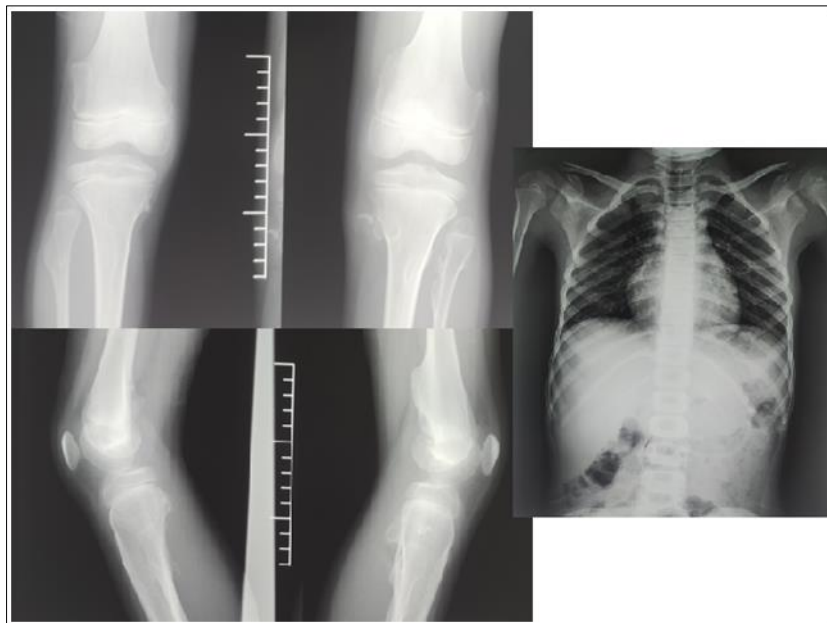
Article History

Received: 06.04.2026

Accepted: 27.05.2026

Published: 02.06.2026

Hereditary multiple exostoses (or multiple osteochondromatosis) is a rare disorder, affecting approximately 1 in 50,000 births. Its transmission is autosomal dominant: a single mutated allele (EXT1 or EXT2 gene) is sufficient to pass on the disease, with high penetrance but highly variable expressivity. The osteocartilaginous outgrowths are electively located at the metaphyses of long bones, which distinguishes them from epiphyseal or vertebral involvement seen in other conditions such as spondyloepiphyseal dysplasia. They present two typical forms – sessile (broad base of implantation) or pedunculated (dome- or club-shaped) – and their direction is centrifugal, growing away from the growth plate toward the outside of the limb, opposite the adjacent joint. The knee radiographs of the 16-year-old boy and his 20-year-old sister perfectly illustrate these features. In the boy, spinal imaging shows no exostosis, confirming the usual sparing of the axial skeleton in this condition.



X-ray of the brother showing both knees and spine

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CITATION: Youness Mokhchani, Bouchaib Chafry, Driss Benchebba, A. S. Bouabid, Mustapha Boussouga (2026). Hereditary Multiple Exostoses: Metaphyseal Location of Outgrowths in Two Siblings (Knees) – Distinction from Spondyloepiphyseal Dysplasia. *South Asian Res J Med Sci*, 8(3): 89-90. 89



X-ray of the sister showing the appearance of the exostoses on the right knee