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Peripheral dysostosis

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Peripheral dysostosis. ORPHA:1795*

Peripheral dysostosis is a rare primary bone dysplasia characterized by cone-shaped epiphyses of the phalanges, hyperextensibility and hyperflexibility of the fingers and marked delay in ossification of hand bones. Short-limbed short stature, very stubby, short fingers and toes, flat face and nose and a large skull may also be associated. There have been no further descriptions in the literature since 1980.