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Metaphyseal acroscyphodysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Metaphyseal acroscyphodysplasia. ORPHA:1240

Metaphyseal acroscyphodysplasia is an extremely rare form of metaphyseal dysplasia characterized by the distinctive radiological sign of cone-shaped upper tibial and lower femoral epiphyses embedded in large cup-shaped metaphyses, associated with short stature and micromelia. Upper limb involvement includes brachydactyly and phalangeal and metacarpal cone-shaped epiphyses. The association of metaphyseal acroscyphodysplasia with psychomotor delay and alopecia has also been reported in some cases.