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Thoracomelic dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Thoracomelic dysplasia. ORPHA:1803

Thoracomelic dysplasia is an extremely rare primary bone dysplasia disorder characterized by a bell-shaped thorax, disproportionate short stature, pelvic hypoplasia, dislocatable radial heads and elongated distal fibulae. No acetabular spurs nor phalangeal cone-shaped epiphyses are present and osseous manifestations tend to normalize with age. There have been no further descriptions in the literature since 1988.

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