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Spondylometaphyseal dysplasia, A4 type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spondylometaphyseal dysplasia, A4 type. ORPHA:168555

Spondylometaphyseal dysplasia, A4 type is a rare primary bone dysplasia disorder characterized by disproportionate short stature, severe femoral neck deformity, marked metaphyseal abnormalities and platyspondyly consisting of ovoid vertebral bodies that have an anterior tongue-like deformity.