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Spondyloepiphyseal dysplasia, Cantu type

INSERM

Source

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

Spondyloepiphyseal dysplasia, Cantu type. ORPHA:163654

Spondyloepiphyseal dysplasia, Cantu type is an extremely rare type of spondyloepiphyseal dysplasia (see this term) described in about 5 patients to date and characterized by clinical signs including short stature, peculiar facies with blepharophimosis, upward slanted eyes, abundant eyebrows and eyelashes, coarse voice, and short hands and feet (brachymetacarpalia, brachymetatarsalia and brachyphalangia).