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Spondyloepimetaphyseal dysplasia, aggrecan type

INSERM

Source

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

Spondyloepimetaphyseal dysplasia, aggrecan type. ORPHA:171866

Spondyloepimetaphyseal dysplasia, aggrecan type is a new form of skeletal dysplasia characterized by severe short stature, facial dysmorphism and characteristic radiographic findings.