Spondylometaphyseal dysplasia, Schmidt type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spondylometaphyseal dysplasia, Schmidt type. ORPHA:93316

Spondylometaphyseal dysplasia, Schmidt type is characterized by short stature, myopia, ,small pelvis, progressive kypho-scoliosis, wrist deformity, severe genu valgum, short long bones, and severe metaphyseal dysplasia with moderate spinal changes and minimal changes in the hands and feet.