Spondylometaphyseal dysplasia, Czarny-Ratajczak type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spondylometaphyseal dysplasia, Czarny-Ratajczak type. ORPHA:370019

Spondylometaphyseal dysplasia, Czarny-Ratajczak type is a rare primary bone dysplasia disorder characterized by short stature with severe shortening of limbs, genu vara deformity and enlarged joints with movement limitation particularly affecting the hip joints. Radiological findings show coxa vara, generalized metaphyseal irregularities of the tubular bones (including cupping, fraying and splaying) which is more severe in the femur and forearm bones than the metacarpals and phalanges, and vertebral abnormalities including ovoid vertebral bodies with anterior rectangular protrusions, and severe platyspondyly.