Spondyloepimetaphyseal dysplasia congenita, Strudwick type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spondyloepimetaphyseal dysplasia congenita, Strudwick type. ORPHA:93346

Spondyloepimetaphyseal dysplasia congenita, Strudwick type is characterized by disproportionate short stature from birth (with a very short trunk and shortened limbs) and skeletal abnormalities (lordosis, scoliosis, flattened vertebrae, pectus carinatum, coxa vara, clubfoot, and abnormal epiphyses or metaphyses).