Chondrodysplasia punctata, tibial-metacarpal type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Chondrodysplasia punctata, tibial-metacarpal type. ORPHA:79346

A rare, non-rhizomelic, chondrodysplasia punctata syndrome characterized, radiologically, by stippled calcifications and disproportionate, short metacarpals and tibiae (with characteristic overshoot of the proximal fibula), clincially manifesting with severe short stature, bilateral shortening of upper and lower limbs, flat midface and nose, in the absence of cataracts and cutaneous anomalies. Neonatal tachnypnea, hydrocephalus and mild developmental delay have been seldomly associated. Additional radiologic features include bowed long bones, platyspondyly and/or vertebral clefts.