Acromesomelic dysplasia, Grebe type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acromesomelic dysplasia, Grebe type. ORPHA:2098

Acromesomelic dysplasia, Grebe type is an autosomal recessively inherited form of acromesomelic dysplasia (see this term) characterized by severe dwarfism at birth, abnormalities confined to limbs, severe shortening and deformity of long bones, fusion or absence of carpal and tarsal bones, ball shaped fingers and, occasionally, polydactyly and absent joints. As seen in acromesomelic dysplasia, Hunter-Thomson type and acromesomelic dysplasia, Maroteaux Type (see these terms), facial features and intelligence are normal.