Acromesomelic dysplasia, Hunter-Thompson type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acromesomelic dysplasia, Hunter-Thompson type. ORPHA:968

Acromesomelic dysplasia, Hunter-Thomson type is an autosomal recessively inherited form of acromesomelic dysplasia (see this term) characterized by severe dwarfism (adult height approximately 120 cm) with abnormalities limited to the limbs (affecting the lower limbs more than upper limbs, with middle and distal segments being the most affected), severe shortening, absence or fusion of tubular bones of hands and feet and large joint dislocations. As seen in acromesomelic dysplasia, Grebe type and acromesomelic dysplasia, Maroteaux type (see these terms), facial features and intelligence are normal.