Omodysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Omodysplasia. ORPHA:2733

Omodysplasia is a rare skeletal dysplasia characterized by severe limb shortening and facial dysmorphism. Two types of omodysplasia have been described: an autosomal recessive or generalized form (also referred to as micromelic dysplasia with dislocation of radius) marked by severe micromelic dwarfism with predominantly rhizomelic shortening of both the upper and lower limbs, and an autosomal dominant form in which stature is normal and shortening is limited to the upper limbs.