Achondrogenesis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Achondrogenesis. ORPHA:932

Achondrogenesis describes a rare group of lethal skeletal dysplasias characterized by an endochondral ossification deficiency that leads to dwarfism with extreme micromelia, a small thorax, a prominent abdomen, anasarca and polyhydramnios. There are three types of achondrogenesis that exist and that differ clinically, radiologically, histologically and genetically: achondrogensis type 1a, type 1b and type 2 (see these terms).