Achondrogenesis type 1B

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Achondrogenesis type 1B. ORPHA:93298

Achondrogenesis type 1B (ACG1B), a form of achondrogenesis (see this term), is a rare lethal skeletal dysplasia characterized by severe micromelia with very short fingers and toes, a flat face, a short neck, thickened soft tissue around the neck, hypoplasia of the thorax, protuberant abdomen, a hydropic fetal appearance and distinctive histological features of the cartilage.