Lethal recessive chondrodysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Lethal recessive chondrodysplasia. ORPHA:1423

Lethal recessive chondrodysplasia is an extremely rare lethal form of chondrodysplasia characterized by severe micromelic dwarfism, short and incurved limbs with normal hands and feet, facial dysmorphism (disproportionately large skull, frontal prominence, slightly flattened nasal bridge and short neck), muscular hypotonia, hyperlaxity of the extremities, and a narrow thorax. Most patients die of respiratory distress during the first hours or weeks of life. There have been no further descriptions in the literature since 1988.