Campomelic dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Campomelic dysplasia. ORPHA:140

Campomelic dysplasia is a very rare disorder characterised by a variable association of skeletal abnormalities (bowed and fragile long bones, pelvis and chest abnormalities, eleven rib pairs instead of the usual twelve), and extraskeletal abnormalities (facial dysmorphology, cleft palate, sexual ambiguity or sex reversal in two thirds of the affected boys, and brain, heart and kidney malformations).