Open Peer Review on Qeios

Dyssegmental dysplasia, Silverman-Handmaker type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dyssegmental dysplasia, Silverman-Handmaker type. ORPHA:1865

Dyssegmental dysplasia, Silverman-Handmaker type is a rare, genetic, primary bone dysplasia disorder, and lethal form of neonatal short-limbed dwarfism, characterized by anisospondyly, severe short stature and limb shortening, metaphyseal flaring and distinct dysmorphic features (i.e. flat facial appearance, abnormal ears, short neck, narrow thorax). Additional features may include other skeletal findings (e.g. joint contractures, bowed limbs, talipes equinovarus) and urogenital and cardiovascular abnormalities.