Axial spondylometaphyseal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Axial spondylometaphyseal dysplasia. ORPHA:168549

Axial spondylometaphyseal dysplasia is a rare type of spondylometaphyseal dysplasia characterized by metaphyseal changes of the truncal-juxtatruncal bones associated with retinal dystrophy. Patients typically present progressive postnatal growth failure with rhizomelic shortening of the limbs, a deformed, hypoplastic thorax and retinitis pigmentosa or pigmentary retinal degeneration. Radiographic findings include short ribs with flared, cupped anterior ends, mild platyspondyly, lacy ilia and metaphyseal dysplasia of the proximal femora.