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Short stature-advanced bone age-early-onset osteoarthritis syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Short stature-advanced bone age-early-onset osteoarthritis syndrome. ORPHA:435804*

A rare, primary bone dysplasia characterized by proportional short stature, early cessation of bone growth, accelerated skeletal maturation, variable presence of early-onset osteoarthritis and osteochondritis dissecans, and normal endocrine evaluation. The variable dysmorphic features include mild to relative macrocephaly, frontal bossing, midfacial hypoplasia, flat nasal bridge, brachydactyly, broad thumbs, and lordosis.