Spondyloenchondrodysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spondyloenchondrodysplasia. ORPHA:1855

Spondyloenchondrodysplasia (SPENCD) is a very rare genetic skeletal dysplasia characterized clinically by skeletal anomalies (short stature, platyspondyly, short broad ilia) and enchondromas in the long bones or pelvis. SPENCD may have a heterogeneous clinical spectrum with neurological involvement (spasticity, mental retardation and cerebral calcifications) or autoimmune manifestations, such as immune thrombocytopenic purpura, systemic lupus erythematosus (see these terms) hemolytic anemia and thyroiditis.