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Pelvic dysplasia-arthrogryposis of lower limbs syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pelvic dysplasia-arthrogryposis of lower limbs syndrome. ORPHA:2840

Pelvic dysplasia-arthrogryposis of lower limbs syndrome is a rare, genetic, dysostosis syndrome characterized by intrauterine growth restriction, short stature (with short lower segment), lower limb joint contractures and muscular hypotrophy, narrow, small pelvis, lumbar hyperlordosis with scoliosis, and foot deformity (short, overlapping toes). Imaging reveals ovoid/wedge-shaped vertebral bodies, pelvic and skeletal hypoplasia with metatarsal fusion in the lower limbs, and normal skull and upper limbs.