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Autosomal dominant congenital benign spinal muscular atrophy

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Autosomal dominant congenital benign spinal muscular atrophy. ORPHA:1216*

Autosomal dominant congenital benign spinal muscular atrophy is a rare distal hereditary motor neuropathy, with a variable clinical phenotype, typically characterized by congenital, non-progressive, predominantly distal, lower limb muscle weakness and atrophy and congenital (or early-onset) flexion contractures of the hip, knee and ankle joints. Reduced or absent lower limb deep tendon reflexes, skeletal anomalies (bilateral talipes equinovarus, scoliosis, kyphoscoliosis, lumbar hyperlordosis), late ambulation, waddling gait, joint hyperlaxity and/or bladder and bowel dysfunction are usually also associated.