Autosomal recessive limb-girdle muscular dystrophy type 2S

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2S. ORPHA:369840

Autosomal recessive limb-girdle muscular dystrophy type 2S (LGMD2S) is a form of limb-girdle muscular dystrophy characterized by childhood-onset of progressive proximal muscle weakness (leading to reduced ambulation) with myalgia and fatigue, in addition to infantile hyperkinetic movements, truncal ataxia, and intellectual disability. Additional manifestations include scoliosis, hip dysplasia, and less commonly, ocular features (e.g. myopia, cataract) and seizures.