Autosomal recessive limb-girdle muscular dystrophy type 2D

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2D. ORPHA:62

Autosomal recessive limb-girdle muscular dystrophy type 2D (LGMD2D) is a subtype of autosomal recessive limb-girdle muscular dystrophy characterized by childhood onset of progressive proximal weakness of the shoulder and pelvic girdle muscles, resulting in difficulty walking, scapular winging, calf hypertrophy and contractures of the Achilles tendon, which lead to a tiptoe gait pattern. Cardiac and respiratory involvement is rare.