Autosomal recessive limb-girdle muscular dystrophy type 2B

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2B. ORPHA:268

Autosomal recessive limb-girdle muscular dystrophy type 2B (LGMD2B) is a subtype of autosomal recessive limb-girdle muscular dystrophy characterized by an onset in late adolescence or early adulthood of slowly progressive, proximal weakness and atrophy of shoulder and pelvic girdle muscles. Cardiac and respiratory muscles are not involved. Hypertrophy of the calf muscles and highly elevated serum creatine kinase levels are frequently observed.