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Autosomal recessive limb-girdle muscular dystrophy type 2F

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2F. ORPHA:219

Autosomal recessive limb-girdle muscular dystrophy type 2F (LGMD2F) is a subtype of autosomal recessive limb-girdle muscular dystrophy characterized by a variable age of onset of progressive weakness and wasting of the proximal skeletal muscles of the shoulder and pelvic girdles, frequently associated with progressive respiratory muscle impairment and cardiomyopathy. Calf hypertrophy, muscle cramps and elevated serum creatine kinase levels are also observed. Neuropsychomotor development is usually normal.