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Autosomal recessive limb-girdle muscular dystrophy type 2X

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2X. ORPHA:476084

Autosomal recessive limb-girdle muscular dystrophy type 2X is a rare subtype of autosomal recessive limb-girdle muscular dystrophy characterized by atrioventricular block resulting in repeated syncope episodes, elevated creatine kinase serum levels and adult-onset of slowly progressive proximal limb skeletal muscle weakness and atrophy. Muscular dystrophic changes observed in muscle biopsy include diameter variability, increased central nuclei, and presence of necrotic and regenerating fibers.