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Autosomal recessive limb-girdle muscular dystrophy type 2Q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2Q. ORPHA:254361

Autosomal recessive limb-girdle muscular dystrophy type 2Q (LGMD2Q) is a form of limb-girdle muscular dystrophy characterized by proximal muscle weakness presenting in early childhood (with occasional falls and difficulties in climbing stairs) and a progressive course resulting in loss of ambulation in early adulthood. Muscle atrophy and multiple contractures have also been reported in rare cases.