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# Autosomal recessive limb-girdle muscular dystrophy type 2U

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2U. ORPHA:352479*

A rare subtype of autosomal recessive limb-girdle muscular dystrophy disorder characterized by infantile to childhood-onset of slowly progressive, principally proximal, shoulder and/or pelvic-girdle muscular weakness that typically presents with positive Gowers' sign and is associated with elevated creatine kinase levels, hyporeflexia, joint and achilles tendon contractures, and muscle hypertrophy, usually of the thighs, calves and/or tongue. Other highly variable features include cerebellar, cardiac and ocular abnormalities.