Autosomal recessive limb-girdle muscular dystrophy type 2Y

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2Y. ORPHA:424261

Autosomal recessive limb-girdle muscular dystrophy type 2Y is a form of limb-girdle muscular dystrophy, presenting in the first or second decades of life, characterized by slowly progressive proximal and distal muscle weakness and atrophy. Additional manifestations include contractures of the proximal and distal interphalangeal hand joints, rigid spine, restricted pulmonary function, and mild cardiomyopathy.