Autosomal recessive limb-girdle muscular dystrophy type 2H

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2H. ORPHA:1878

Autosomal recessive limb-girdle muscular dystrophy type 2H (LGMD2H) is a mild subtype of autosomal recessive limb girdle muscular dystrophy characterized by slowly progressive proximal muscle weakness and wasting of the pelvic and shoulder girdles with onset that usually occurs during the second or third decade of life. Clinical presentation is variable and can include calf psuedohypertrophy, joint contractures, scapular winging, muscle cramping and/or facial and respiratory muscle involvement.