Autosomal dominant limb-girdle muscular dystrophy type 1H

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant limb-girdle muscular dystrophy type 1H. ORPHA:238755

Autosomal dominant limb-girdle muscular dystrophy type 1H (LGMD1H) is a subtype of autosomal dominant limb-girdle muscular dystrophy characterized by slowly progressive proximal muscular weakness initially affecting the lower limbs (and later involving the upper limbs), hypotrophy of upper and lower limb-girdle muscles, hyporeflexia, calf hypertrophy, and increased serum creatine kinase. There is no involvement of oculo-facial-bulbar muscles and cardiac muscle.