Autosomal recessive limb-girdle muscular dystrophy type 2E

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2E. ORPHA:119

Autosomal recessive limb girdle muscular dystrophy type 2E (LGMD2E) is a subtype of autosomal recessive limb girdle muscular dystrophy characterized by a childhood to adolescent onset of progressive pelvic- and shoulder-girdle muscle weakness, particularly affecting the pelvic girdle (adductors and flexors of hip). Usually the knees are the earliest and most affected muscles. In advanced stages, involvement of the shoulder girdle (resulting in scapular winging) and the distal muscle groups are observed. Calf hypertrophy, cardiomyopathy, respiratory impairment, tendon contractures, scoliosis, and exercise-induced myoglobinuria may be observed.