Autosomal dominant limb-girdle muscular dystrophy type 1A

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant limb-girdle muscular dystrophy type 1A. ORPHA:266

Autosomal dominant limb-girdle muscular dystrophy type 1A (LGMD1A) is a subtype of autosomal dominant limb girdle muscular dystrophy characterized by an adult onset of proximal shoulder and hip girdle weakness (that later progresses to include distal weakness), nasal speech and dysarthria. Other frequent findings include tightened heel cords, reduced deep-tendon reflexes and elevated creatine kinase serum levels. Respiratory failure, as well as mild facial weakness and dysphagia, may also be observed.