Autosomal dominant limb-girdle muscular dystrophy type 1F

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant limb-girdle muscular dystrophy type 1F. ORPHA:55595

Autosomal dominant limb-girdle muscular dystrophy type 1F (LGMD1F) is a subtype of autosomal dominant limb-girdle muscular dystrophy ,with a variable age of onset, characterized by progressive, proximal weakness and wasting of the shoulder and pelvic musculature (with the pelvic girdle, and especially the ileopsoas muscle, being more affected) and frequent association of calf hypertrophy, dysphagia, arachnodactyly with or without finger contractures and/or distal and axial muscle involvement. Additional features include an abnormal gait, exercise intolerance, myalgia, fatigue and respiratory insufficiency. Cardiac conduction defects are typically not observed.