Autosomal recessive limb-girdle muscular dystrophy type 2W

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive limb-girdle muscular dystrophy type 2W. ORPHA:466801

Autosomal recessive limb-girdle muscular dystrophy type 2W is a subtype of autosomal recessive limb girdle muscular dystrophy characterized by childhood onset of severe, progressive, proximal skeletal muscle weakness and atrophy of the upper and lower limbs with later involvement of distal muscles and development of severe quadraparesis, calf hypertrophy, triangular tongue, and dilated cardiomyopathy. Skeletal muscles undergo diffuse, bilateral, symmetric and severe atrophy with fat infiltration.