TABLE 462e-7 AUTOSOMAL RECESSIVE LIMB-GIRDLE MUSCULAR DYSTROPHIES (LGMDs)

Disease	Clinical Features	Laboratory Features	Abnormal Protein
LGMD2A	Onset first or second decade Scapular winging; no calf hypertrophy; no cardiac or respiratory muscle weakness Proximal and distal weakness; may have contractures at elbows, wrists, and fingers	Serum CK 3–15× normal EMG myopathic Muscle biopsy may show lobulated muscle fibers	Calpain-3
LGMD2B	Onset second or third decade Proximal muscle weakness at onset, later distal (calf) muscles affected Miyoshi's myopathy is variant of LGMD2B with calf muscles affected at onset	Serum CK 3–100× normal EMG myopathic Inflammation on muscle biopsy may simulate polymyositis	Dysferlin
LGMD2C–F	Onset in childhood to teenage years Clinical condition similar to Duchenne and Becker muscular dystrophies Cognitive function normal	Serum CK 5–100× normal EMG myopathic	γ, α, β, δ sarcoglycans
LGMD2G	Onset age 10 to 15 Proximal and distal muscle weakness	Serum CK 3–17× normal EMG myopathic Muscle biopsy may show rimmed vacuoles	Telethonin
LGMD2H	Onset first to third decade Proximal muscle weakness	Serum CK 2–25× normal EMG myopathic	*TRIM32* gene
LGMD2I	Onset first to third decade Clinical condition similar to Duchenne or Becker dystrophies Cardiomyopathy and respiratory failure may occur early before significant weakness Cognitive function normal	Serum CK 10–30× normal EMG myopathic	Fukutin-related protein
LGMD2J[a]	Onset first to third decade Proximal lower limb weakness Mild distal weakness Progressive weakness causes loss of ambulation	Serum CK 1.5–2× normal EMG myopathic Muscle biopsy reveals rimmed vacuoles	Titin
LGMD2K	Usually presents in infancy as Walker-Warburg syndrome but can present in early adult life with proximal weakness and only minor CNS abnormalities	CK 10–20× normal EMG myopathic	POMT1
LGMD2L	Presents in childhood or adult life May manifest with quadriceps atrophy and myalgia Some present with early involvement of the calves in the second decade of life, resembling Miyoshi's myopathy (dysferlinopathy)	CK 8–20× normal EMG myopathic	Anoctamin 5
LGMD2M	Usually presents in infancy as Fukuyama's congenital muscular dystrophy but can present in early adult life with proximal weakness and only minor CNS abnormalities	CK 10–50× normal EMG myopathic	Fukutin
LGMD2N	Usually presents in infancy as muscle-eye-brain disease but can present in early adult life with proximal weakness and only minor CNS abnormalities	CK 5–20× normal EMG myopathic	POMGnT1
LGMD2O	Usually presents in infancy as Walker-Warburg syndrome but can present in early adult life with proximal weakness and only minor CNS abnormalities	CK 5–20× normal EMG myopathic	POMT2
LGMD2P	One case reported presenting in early childhood	CK >10× normal	α-Dystroglycan
LGMD2Q	Onset in infancy to fourth decade; proximal weakness; may have ptosis and extraocular weakness; epidermolysis bullosa (also considered a congenital myasthenic syndrome)	CK variable, but usually only mildly elevated EMG myopathic Repetitive nerve stimulation may show decrement	Plectin 1
LGMD2R	See LGMD1E (Table 462e-6)	See LGMD1E	Desmin
LGMD2S	Onset in infancy to sixth decade Proximal weakness Eye abnormalities common; truncal ataxia and chorea Mild to moderate intellectual disability Hutterite descent	CK 1.5–20× normal	TRAPC11

[a]Udd's type distal myopathy is a form of titin deficiency with only distal muscle weakness (see Table 462e-9).

Abbreviations: CK, creatine kinase; EMG, electromyography; NCS, nerve conduction studies; POMT1, protein-O-mannosyltransferase 1; POMT2, protein-O-mannosyltransferase 2; POMGNT1, O-linked mannose beta 1,2-N-acetylglucosaminyltransferase; TNPO3, transportin 3; TRAPC11, transport (trafficking) protein particle complex, subunit 11.