E-TABLE 122-1 LIMB-GIRDLE MUSCULAR DYSTROPHIES—ABBREVIATED LIST

INHERITANCE	NOMENCLATURE	MUTATION	AGE	CK	PHENOTYPES	SURVEILLANCE
Autosomal dominant*	1A	5q31; MYOT (myotilin)	18-40	Normal to 15x	limb-girdle pattern; tight Achilles tendons; dysarthria	Respiratory involvement; trouble swallowing
	1B	1q21; LMNA (lamin A/C)	Variable; birth to adulthood	Normal to mildly elevated	Limb-girdle pattern; contractures	Significant cardiac involvement
	1C	3p25 CAV3 (caveolin 3)	5-adulthood	3x-40x	Limb-girdle pattern; rippling muscle disease; elevated CK and cramping; distal myopathy (hand/foot)	Cardiac involvement
	1D	2q35; DES (desmin)	Teens to adulthood	2x-4x	Limb-girdle pattern	Cardiac involvement
	1E	7q36 DNAJB6 (	30s-60s	Normal to 5x	Limb-girdle pattern; lower > upper	Trouble swallowing
Autosomal recessive†	2A	15q15 CAPN3(Calpain-3)	2-40s	Normal to 80x	Limb-girdle pattern; scapular winging; paraspinal involvement/ scoliosis	
	2B	2p13 DYSF (dysferlin)	Teens to 20s	10x to 72x	Limb-girdle pattern; Miyoshi distal myopathy (calf wasting); tibialis anterior	Respiratory late
	2C-2F	13q12 (γ- sarcoglycan); 17q12 (α- sarcoglycan); 4q12 (β -sarcoglycan); 5q33 (δ- sarcoglycan)	3-15	>10x	Limb-girdle pattern; mild to severe phenotype; contractures/scoliosis; calf pseudo-hypertrophy	Respiratory and cardiac involvement; orthopedics
	2G	17q12 TCAP (telethonin)	9-15	3x-30x	Limb-girdle pattern; distal tibialis anterior	
	2I	19q13 FKRP (fukutin related protein)	Birth to 20s	10x to 30x	Limb-girdle pattern; upper > lower	Respiratory involvement

*Other autosomal dominant mutations: 1F 7q32 TNPO3; 1G 4q21; 1H 3p23.
†Other recessive mutations: 2H 9q33 TRIM32; 2J 2q24 Titin; 2L 11p14 ANO5.