

Table 6: Limb-girdle muscular dystrophy: Autosomal Recessive

<u>Disease</u>	<u>Onset</u>	<u>HyperCKemia</u>	<u>Progression</u>	<u>Skeletal Muscular Findings</u>	<u>Extramuscular findings</u>
LGMD R1 calpain3-related	Adolescence	High to very high	Moderate / rapid	Proximal legs / Rectus abdominus / Peri-scapular; contractures	usually no cognitive involvement
LGMD R2 dysferlin-related	Young adulthood	Very high	Slow	Asymmetric weakness; preference for biceps with spared deltoid; Absent or reduced dysferlin staining on muscle biopsy	Respiratory insufficiency with early onset;
LGMD R3 a-sarcoglycan-related	Early childhood	Very high	Rapid	Quadriceps involvement; calf hypertrophy	Cardiomyopathy
LGMD R4 b-sarcoglycan-related	Early childhood	Very high	Rapid	Tongue hypertrophy; Muscle hypertrophy	Cardiomyopathy
LGMD R5 y-sarcoglycan-related	Early childhood	Very high	Moderate / Rapid	Calf hypertrophy; lumbar hyperlordosis; scapular winging; tongue hypertrophy; sparing of quadriceps	Respiratory insufficiency; Hearing loss; Cardiomyopathy later in disease course
LGMD R6 d-sarcoglycan-related	Early childhood	Very high	Rapid	Calf hypertrophy; cramps	Cardiomyopathy
LGMD R7 telethonin-related	Adolescence	High to very high	Slow	Calf hypertrophy or atrophy; normal or weak facial muscles; cramps	Cardiomyopathy
LGMD R8 TRIM32-related	Adulthood	High	Slow	Variable	
LGMD R9 FKRP-related	Late child- to adulthood	Very high	Variable	Facial involvement later in disease; can be asymmetric; exertional pain / cramps limbs; myoglobinuria	Respiratory failure -30%
LGMD R10 titin-related	Young adulthood	High	Rapid	Can be asymmetric; late onset distal weakness possible	
LGMD R11 POMT1-related	Childhood	Very high	Slow	Ankle joint contractures – 50%	Some intellectual disability
LGMD R12 anoctamin5-related	Young to late adulthood	Normal to very high	Slow	Asymmetric; legs > arms; can have facial involvement; normal strength in distal arms;	
LGMD R13 Fukutin-related	Early childhood	Very high	Moderate	Legs > arms; mild to moderate facial involvement	Some intellectual disability
LGMD R14 POMT2-related	Early childhood	High	Slow	Calf hypertrophy; scapular winging; spinal lordosis	Some intellectual disability
LGMD R15 POMGnT1-related	Early to late childhood	Mildly elevated	Moderate	Congenital hypotonia	Myopia; glaucoma; juvenile cataracts; retinal dysplasia; intellectual disability
LGMD R16 a-dystroglycan related	Early childhood	Very High	Slow	Contractures common	Some intellectual disability; small head circumference
LGMD R17 plectin-related	Early childhood	Moderate to high	Moderate	Atrophic musculature and contractures late	
LGMD R18 TRAPPC11-related	Early to late childhood	High	Moderate	Legs>Arms	Movement disorder; Some intellectual disability; seizures
LGMD R19 GMPPB-related	Birth to adulthood	Moderate to high	Slow	Rhabdomyolysis and cramps	Cardiomyopathy-rare; Some intellectual disability
LGMD R20 ISPD-related	<1 year old	Very High	Moderate	Proximal weakness	Myopia; Oculomotor apraxia; cerebellar hypoplasia; Some intellectual disability