

# MUSCLE DISORDERS GENE PANEL DGD141114

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM ID</i>
ACADVL	88.4	100%	97%	VLCAD deficiency, 201475
ACTA1	73.6	99%	94%	Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ACVR1	91.8	100%	97%	Fibrodysplasia ossificans progressiva, 135100
AGL	146.2	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	80.7	97%	90%	Myasthenia, limb-girdle, familial, 254300
ANO5	103.4	100%	100%	Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, type 2L, 611307 Miyoshi muscular dystrophy 3, 613319
ATP2A1	121.2	100%	99%	Brody myopathy, 601003
B3GNT1	106.0	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	151.0	100%	100%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BICD2	97.8	99%	96%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 -3
BIN1	54.9	85%	75%	Myopathy, centronuclear, autosomal recessive, 255200
CACNA1S	96.0	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CAPN3	115.8	99%	97%	Muscular dystrophy, limb-girdle, type 2A, 253600
CASQ1	111.1	100%	100%	No OMIM phenotype
CAV3	146.7	100%	100%	Muscular dystrophy, limb-girdle, type IC, 607801 Rippling muscle disease, 606072 Creatine phosphokinase, elevated serum, 123320 Myopathy, distal, Tateyama type, 614321 Cardiomyopathy, familial hypertrophic, 192600 Long QT syndrome-9, 611818
CCDC78	90.8	100%	100%	Myopathy, centronuclear, 4, 614807

CFL2	133.6	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	69.3	91%	77%	Myasthenic syndrome, congenital, associated with episodic apnea, 254210
CHKB	91.1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	102.3	100%	98%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNA1	102.5	98%	94%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CHRND	97.1	96%	90%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNE	158.8	100%	100%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CLCN1	91.2	100%	97%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive
CNTN1	110.7	100%	99%	Myopathy, congenital, Compton-North, 612540
COL12A1	115.4	100%	99%	{Lung cancer, susceptibility to, association with}(Rudd (2006) Genome Res 16,693) Bethlem-like myopathy (Hicks (2014) Hum Mol Genet 23,2353) Joint hypermobility syndrome with myopathy (Zou (2014) Hum Mol Genet 23, 2339)
COL6A1	81.8	98%	96%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 {Ossification of the posterior longitudinal spinal ligaments}, 602475 (2)
COL6A2	82.7	99%	97%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 Myosclerosis, congenital, 255600
COL6A3	124.3	99%	99%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090
COLQ	68.2	100%	89%	Endplate acetylcholinesterase deficiency, 603034
CPT2	109.4	92%	91%	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212

CRYAB	133.3	100%	100%	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184
DAG1	142.3	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DES	92.8	96%	88%	?Muscular dystrophy, limb-girdle, type 2R, 615325 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DMD	57.4	98%	95%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DNAJB6	43.9	88%	80%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNM2	80.9	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Lethal congenital contracture syndrome 5, 615368
DOK7	53.0	93%	79%	Myasthenia, limb-girdle, familial, 254300 Fetal akinesia deformation sequence, 208150
DPAGT1	90.9	99%	95%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM2	59.9	98%	88%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	101.2	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DYNC1H1	117.4	99%	96%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYSF	97.4	99%	99%	Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768 Miyoshi muscular dystrophy 1, 254130
EMD	81.9	100%	92%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	106.8	99%	96%	Glycogen storage disease XIII, 612932
ERBB3	111.9	100%	99%	Lethal congenital contractural syndrome 2, 607598
FAM111B	183.1	100%	98%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy and pulmonary fibrosis, 615704
FHL1	42.8	96%	77%	Hemophagocytic lymphohistiocytosis, familial, 1 (2)

FKRP	80.2	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	108.0	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNC	94.1	96%	94%	Myopathy, myofibrillar, 5, 609524 Myopathy, distal, 4, 614065 -3
GAA	97.7	100%	98%	Glycogen storage disease II, 232300
GBE1	104.3	98%	94%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	105.9	100%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GENE	99.5	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GYG1	51.0	82%	58%	Glycogen storage disease XV, 613507
GYS1	64.1	95%	81%	Glycogen storage disease 0, muscle, 611556
HSPG2	73.3	98%	93%	Schwartz-Jampel syndrome, type 1, 255800 Dyssegmental dysplasia, Silverman-Handmaker type, 224410
IGHMBP2	70.4	97%	88%	Neuronopathy, distal hereditary motor, type VI, 604320
ISCU	85.5	100%	96%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	88.9	95%	94%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITGA7	86.0	98%	93%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	54.0	100%	96%	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	110.4	97%	92%	Andersen syndrome, 170390 Short QT syndrome-3, 609622 Atrial fibrillation, familial, 9, 613980
KLHL41	147.8	100%	100%	Nemaline myopathy 9, 615731
KLHL9	157.1	100%	100%	No OMIM phenotype Myopathy, distal, early-onset (Cirak (2010) Brain 133, 2123)

LAMA2	102.7	100%	98%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMP2	61.4	98%	90%	Danon disease, 300257
LARGE	105.4	99%	95%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LDB3	86.0	94%	93%	Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493
LDHA	46.2	85%	67%	Glycogen storage disease XI, 612933
LMNA	72.2	97%	89%	Emery-Dreifuss muscular dystrophy 2, AD, 181350 Cardiomyopathy, dilated, 1A, 115200 Lipodystrophy, familial partial, 2, 151660 Emery-Dreifuss muscular dystrophy 3, AR, 181350 Charcot-Marie-Tooth disease, type 2B1, 605588 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Mandibuloacral dysplasia, 248370 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Heart-hand syndrome, Slovenian type, 610140 Malouf syndrome, 212112
LMOD3	186.7	100%	100%	Nemaline myopathy 10, 616165
LPIN1	100.3	100%	99%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MATR3	119.3	97%	95%	Myopathy, distal 2, 606070
MEGF10	101.9	100%	98%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MICU1	97.0	100%	98%	Myopathy with extrapyramidal signs, 615673
MSTN	196.3	100%	100%	Muscle hypertrophy, 614160
MTM1	55.5	100%	98%	Myotubular myopathy, X-linked, 310400
MUSK	130.9	100%	98%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
MYBPC3	90.7	95%	94%	Cardiomyopathy, familial hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396

MYF6	145.5	100%	100%	Myopathy, centronuclear, 3, 614408
MYH2	100.8	97%	93%	Inclusion body myopathy-3, 605637
MYH3	111.2	97%	95%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680
MYH7	89.3	95%	88%	Cardiomyopathy, familial hypertrophic, 1, 192600 Cardiomyopathy, dilated, 1S, 613426 Myopathy, myosin storage, 608358 Laing distal myopathy, 160500 Scapuloperoneal syndrome, myopathic type, 181430 Left ventricular noncompaction 5, 613426
MYOT	140.1	100%	100%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOT	140.1	100%	100%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
NEB	88.9	82%	80%	Nemaline myopathy 2, autosomal recessive, 256030
PABPN1	55.9	67%	54%	Oculopharyngeal muscular dystrophy, 164300
PFKM	103.2	100%	100%	Glycogen storage disease VII, 232800
PGAM2	96.8	100%	100%	Glycogen storage disease X, 261670
PGK1	42.7	75%	67%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	109.2	100%	99%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHKA1	48.2	96%	90%	? Muscle glycogenosis, 300559
PIP5K1C	60.8	91%	87%	Lethal congenital contractural syndrome 3, 611369
PLEC	97.8	99%	96%	Muscular dystrophy with epidermolysis bullosa simplex, 226670 Epidermolysis bullosa simplex, Ogna type, 131950 Epidermolysis bullosa simplex with pyloric atresia, 612138 Muscular dystrophy, limb-girdle, type 2Q, 613723
PNPLA2	78.5	100%	91%	Neutral lipid storage disease with myopathy, 610717
POMGNT1	96.9	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157

POMK	128.9	100%	100%	?Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	104.0	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	74.5	100%	93%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PTRF	151.4	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
PYGM	97.3	100%	98%	McArdle disease, 232600
RAPSN	87.0	91%	84%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency, 608931 Fetal akinesia deformation sequence, 208150
RYR1	80.9	97%	92%	{Malignant hyperthermia susceptibility 1}, 145600 Central core disease, 117000 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 King-Denborough syndrome, 145600
SCN4A	132.7	100%	99%	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, acetazolamide-responsive, 614198 Hypokalemic periodic paralysis, type 2, 613345
SEPN1	74.2	87%	77%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	93.4	94%	87%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	138.7	96%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	94.7	100%	100%	Muscular dystrophy, limb-girdle, type 2F, 601287 Cardiomyopathy, dilated, 1L, 606685
SGCG	105.5	100%	100%	Muscular dystrophy, limb-girdle, type 2C, 253700

SLC52A2	116.3	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SPEG	80.1	98%	89%	Centronuclear myopathy 5, 615959
STIM1	85.9	97%	94%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
SYNE1	110.6	99%	97%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE2	109.3	100%	98%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
TCAP	38.8	68%	48%	Muscular dystrophy, limb-girdle, type 2G, 601954 Cardiomyopathy, dilated, 1N, 607487
TMEM5	168.4	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TNNI2	76.2	100%	98%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNT1	91.4	96%	92%	Nemaline myopathy 5, Amish type, 605355
TPM2	99.8	100%	100%	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	90.1	100%	97%	Nemaline myopathy 1, autosomal dominant or recessive, 609284 CAP myopathy 1, 609284 Myopathy congenital, with fiber-type disproportion, 255310
TRAPPC11	116.1	100%	99%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRIM32	106.2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TRPV4	93.1	100%	99%	Brachyolmia type 3, 113500 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Metatropic dysplasia, 156530 Hereditary motor and sensory neuropathy, type IIc, 606071 Scapuloperoneal spinal muscular atrophy, 181405 [Sodium serum level QTL 1], 613508 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Digital arthropathy-brachydactyly, familial, 606835
TTC19	72.4	87%	77%	Mitochondrial complex III deficiency, nuclear type 2, 615157



TTN	152.5	99%	99%	Cardiomyopathy, familial hypertrophic, 9, 613765 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Myopathy, early-onset, with fatal cardiomyopathy, 611705
UBA1	66.0	100%	98%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	111.2	99%	96%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VIPAS39	115.3	100%	97%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404

*Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014*

*Median Coverage describes the average number of reads seen across 50 exomes*

*% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x*

*% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x*

*OMIM release used for OMIM disease identifiers and descriptions : 31 october 2014*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*