MUSCLE DISORDERS GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ACADVL	107,5	100%	98%	VLCAD deficiency, 201475
ACTA1	89,5	99%	96%	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	112,7	100%	100%	Fibrodysplasia ossificans progressiva, 135100
AGL	155,5	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	108,9	99%	95%	Myasthenia, limb-girdle, familial, 254300
ANO5	114,4	100%	100%	Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, type 2L, 611307 Miyoshi muscular dystrophy 3, 613319
ATP2A1	143,7	100%	100%	Brody myopathy, 601003
B3GNT1	124	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	190,7	100%	100%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BICD2	116,8	98%	95%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 -3
BIN1	64,5	90%	82%	Myopathy, centronuclear, autosomal recessive, 255200
CACNA1S	112,7	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CAPN3	131,9	97%	97%	Muscular dystrophy, limb-girdle, type 2A, 253600

CAV3	194,2	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	125,7	100%	100%	Myopathy, centronuclear, 4, 614807
CFL2	139,8	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	87,5	90%	82%	Myasthenic syndrome, congenital, associated with episodic apnea, 254210
CHKB	107,2	99%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	118,8	100%	98%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	111,8	99%	95%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CHRND	118,6	100%	98%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNE	197,2	100%	100%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CLCN1	103,4	100%	100%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive
CNTN1	128,4	100%	100%	Myopathy, congenital, Compton-North, 612540
COL12A1	121,7	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	110,5	100%	98%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 {Ossification of the posterior longitudinal spinal ligaments}, 602475 (2)
COL6A2	107,7	100%	98%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 Myosclerosis, congenital, 255600
COL6A3	141,6	99%	99%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090
COLQ	84,4	100%	99%	Endplate acetylcholinesterase deficiency, 603034
CPT2	133,7	99%	92%	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	147,7	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	154	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DES	125,2	97%	93%	?Muscular dystrophy, limb-girdle, type 2R, 615325 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DMD	62	98%	94%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DNAJB6	51,8	92%	83%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNM2	91,4	100%	98%	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	74,8	95%	88%	Myasthenia, limb-girdle, familial, 254300 Fetal akinesia deformation sequence, 208150

DPAGT1	109,8	100%	97%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM2	83	99%	98%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	119	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DYNC1H1	136,1	99%	98%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYSF	113,4	100%	100%	Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768 Miyoshi muscular dystrophy 1, 254130
EMD	107,3	100%	93%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	122,8	99%	97%	Glycogen storage disease XIII, 612932
ERBB3	131,8	100%	100%	Lethal congenital contractural syndrome 2, 607598
FHL1	43	93%	76%	Hemophagocytic lymphohistiocytosis, familial, 1 (2)
FKRP	109,7	100%	99%	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	130,9	100%	100%	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	119,3	96%	96%	Myopathy, myofibrillar, 5, 609524 Myopathy, distal, 4, 614065 -3
GAA	131,9	100%	100%	Glycogen storage disease II, 232300
GBE1	122,8	100%	96%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	122,7	99%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542

GNE	109,1	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GYG1	58,1	87%	58%	Glycogen storage disease XV, 613507
GYS1	82	97%	91%	Glycogen storage disease 0, muscle, 611556
HSPG2	95,5	99%	97%	Schwartz-Jampel syndrome, type 1, 255800 Dyssegmental dysplasia, Silverman-Handmaker type, 224410
IGHMBP2	90,4	99%	94%	Neuronopathy, distal hereditary motor, type VI, 604320
ISCU	105,1	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	108,3	96%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITGA7	111,6	99%	95%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	68,8	100%	98%	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	144,7	95%	91%	Andersen syndrome, 170390 Short QT syndrome-3, 609622 Atrial fibrillation, familial, 9, 613980
KLHL9	161	100%	100%	No OMIM phenotype Myopathy, distal, early-onset (Cirak (2010) Brain 133, 2123)
LAMA2	115,9	100%	99%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMP2	69,3	99%	96%	Danon disease, 300257
LARGE	117,1	99%	97%	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	110,2	95%	94%	Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493
LDHA	42,3	76%	65%	Glycogen storage disease XI, 612933

LMNA	86,3	96%	90%	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
LPIN1	121,5	100%	100%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MATR3	134,9	98%	94%	Myopathy, distal 2, 606070
MEGF10	117	100%	99%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MSTN	190,2	100%	100%	Muscle hypertrophy, 614160
MTM1	60,9	100%	99%	Myotubular myopathy, X-linked, 310400
MUSK	149,9	99%	98%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
MYBPC3	116,2	99%	96%	Cardiomyopathy, familial hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYF6	174,8	100%	100%	Myopathy, centronuclear, 3, 614408
MYH2	114,5	99%	94%	Inclusion body myopathy-3, 605637
MYH3	131,9	99%	96%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680

MYH7	105,1	97%	93%	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	152,4	100%	100%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
NEB	98,8	82%	80%	Nemaline myopathy 2, autosomal recessive, 256030
PABPN1	69,5	72%	65%	Oculopharyngeal muscular dystrophy, 164300
PFKM	115,6	100%	99%	Glycogen storage disease VII, 232800
PGAM2	120,2	100%	100%	Glycogen storage disease X, 261670
PGK1	48,6	81%	71%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	120,1	100%	99%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHKA1	54,9	96%	91%	? Muscle glycogenosis, 300559
PIP5K1C	78,8	95%	90%	Lethal congenital contractural syndrome 3, 611369
PLEC	131,4	100%	98%	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	101,3	100%	99%	Neutral lipid storage disease with myopathy, 610717
POMGNT1	113,7	100%	100%	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
POMT1	118,4	100%	99%	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	86,9	99%	95%	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	191,4	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
PYGM	116,7	100%	99%	McArdle disease, 232600
RAPSN	117	91%	86%	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	94,5	98%	95%	{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	157,1	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	97,3	89%	84%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	113,7	97%	91%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	133,7	96%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	119,8	100%	100%	Muscular dystrophy, limb-girdle, type 2F, 601287 Cardiomyopathy, dilated, 1L, 606685
SGCG	110,9	100%	97%	Muscular dystrophy, limb-girdle, type 2C, 253700
SLC52A2	136,4	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SYNE1	126,4	99%	98%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998

SYNE2	124,5	100%	99%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
TCAP	41,8	84%	58%	Muscular dystrophy, limb-girdle, type 2G, 601954 Cardiomyopathy, dilated, 1N, 607487
TMEM5	195,2	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TNNI2	106,3	100%	99%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNT1	108,7	99%	92%	Nemaline myopathy 5, Amish type, 605355
TPM2	118,1	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	87,9	100%	94%	Nemaline myopathy 1, autosomal dominant or recessive, 609284 CAP myopathy 1, 609284 Myopathy congenital, with fiber-type disproportion, 255310
TRAPPC11	131,2	100%	100%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRIM32	123	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TRPV4	115,6	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	82	92%	84%	Mitochondrial complex III deficiency, nuclear type 2, 615157

TTN	168,6	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	73,4	99%	97%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	130,2	100%	97%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VIPAS39	130,2	99%	97%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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 : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors