MUSCLE DISORDERS GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ACADVL	87.7	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	146.4	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
B3GALNT2	83.7	91%	89%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,11,615181
B3GNT1	106	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	151	100%	100%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BICD2	96.2	99%	96%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 -3
BIN1	53.5	85%	74%	Myopathy, centronuclear, autosomal recessive, 255200
CACNA1S	96	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CAPN3	112.5	99%	97%	Muscular dystrophy, limb-girdle, type 2A, 253600
CASQ1	111.1	100%	100%	Myopathy,vacuolar,with CASQ1 aggregates,616231
CAV3	144.9	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	66.4	90%	74%	Myasthenic syndrome, congenital, associated with episodic apnea, 254210
CHCHD10	22.5	66%	28%	?Myopathy,isolated mitochondrial,autosomal dominant,616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2,615911 Spinal muscular atrophy,Jokela type, 615048
CHKB	91.1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	104.6	100%	98%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 60893 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	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	109	100%	99%	Myopathy, congenital, Compton-North, 612540
COL12A1	115.5	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	99%	99%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090

COLQ	70	100%	90%	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	139.7	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	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	57.5	98%	95%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DNAJB6	42.4	85%	78%	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	53.8	90%	76%	Myasthenia, limb-girdle, familial, 254300 Fetal akinesia deformation sequence, 208150
DPAGT1	96.6	99%	95%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM2	70.8	99%	93%	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	98.8	99%	99%	Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768 Miyoshi muscular dystrophy 1, 254130

ECEL1	62	96%	79%	Arthrogryposis,distal,type 5D,615065
EMD	84.8	100%	95%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	106.8	99%	96%	Glycogen storage disease XIII, 612932
ERBB3	115.1	100%	99%	Lethal congenital contractural syndrome 2, 607598
EXOSC8	69.2	98%	92%	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	183.1	100%	98%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy and pulmonary fibrosis, 615704
FHL1	43.9	95%	78%	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 (with or without mental retardation), type B, 5, 606612
FKTN	108.3	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
FLNC	94.1	96%	94%	Myopathy, myofibrillar, 5, 609524 Myopathy, distal, 4, 614065
GAA	97.7	100%	98%	Glycogen storage disease II, 232300
GBE1	104.6	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
GNE	98.4	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GYG1	50.3	83%	60%	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	81	100%	95%	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	85.7	98%	94%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	54	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	105.2	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	94%	93%	Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493
LDHA	50.4	80%	64%	Glycogen storage disease XI, 612933
LMNA	72.5	97%	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
LMOD3	186.7	100%	100%	Nemaline myopathy 10, 616165
LPIN1	99.2	100%	99%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MATR3	124.4	97%	95%	Myopathy, distal 2, 606070
MEGF10	101.8	100%	98%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MICU1	95.2	100%	98%	Myopathy with extrapyramidal signs, 615673
MSTN	196.3	100%	100%	Muscle hypertrophy, 614160
MTM1	54	100%	96%	Myotubular myopathy, X-linked, 310400
MUSK	119.8	100%	98%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
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
NEB	88.9	82%	80%	Nemaline myopathy 2, autosomal recessive, 256030
ORAI1	79.1	99%	87%	Immunodeficiency 9,612782 Myopathy,tubular aggregate, 2,615883
PABPN1	57.2	67%	55%	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	57	85%	81%	Lethal congenital contractural syndrome 3, 611369
PLEC	97.5	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	98.3	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151
POMGNT2	135.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,8),614830
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	103.9	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
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
PTRF	151.4	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
PYGM	92.4	100%	98%	McArdle disease, 232600

RAPSN	83.3	93%	88%	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
RBCK1	80.8	90%	90%	Polyglucosan body myopathy 1 with or without immunodeficiency,615895
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	132.6	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	87%	77%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	91.5	95%	88%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	138.7	96%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	94.9	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
SLC52A3	69.1	100%	95%	Brown-Vialetto-Van Laere syndrome 1,211530 Fazio-Londe disease,211500
SPEG	79.8	97%	88%	Centronuclear myopathy 5, 615959
STIM1	87.9	98%	95%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
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	73.8	100%	97%	Arthrogryposis multiplex congenita, distal, type 2B, 601680

TNNT1	91.4	96%	92%	Nemaline myopathy 5, Amish type, 605355
TNPO3	93.6	100%	98%	Muscular dystrophy,limb-girdle,type 1F,608423
TPM2	90.5	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	68.3	82%	80%	Nemaline myopathy 1, autosomal dominant or recessive, 609284 CAP myopathy 1, 609284 Myopathy congenital, with fiber-type disproportion, 255310
TRAPPC11	116.6	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	94.3	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
TTC19	70.2	90%	83%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	149.2	98%	97%	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
UBA1	66.6	100%	97%	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	111.2	100%	97%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VRK1	133.6	100%	100%	Pontocerebellar hypoplasia type 1A,607596

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
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 : November 15th, 2015

This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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