MUSCLE DISORDERS GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ACADVL	107.6	99%	96%	VLCAD deficiency, 201475
ACTA1	92.7	99%	92%	?Myopathy,scapulohumeroperoneal,616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACVR1	169	100%	100%	Fibrodysplasia ossificans progressiva, 135100
AGL	147.4	100%	99%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	99	93%	89%	Myasthenia, limb-girdle, familial, 254300
ANO5	142.2	97%	94%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ATP2A1	150.9	100%	100%	Brody myopathy, 601003
B3GALNT2	115.4	90%	88%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,11,615181
B3GNT1	101.9	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	94.9	100%	99%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BICD2	126.8	100%	99%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 -3
BIN1	92.7	99%	93%	Myopathy, centronuclear, autosomal recessive, 255200
CACNA1S	132.8	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CAPN3	113.1	98%	93%	Muscular dystrophy, limb-girdle, type 2A, 253600
CASQ1	119.4	100%	100%	Myopathy,vacuolar,with CASQ1 aggregates,616231

CAV3	269.3	100%	100%	Cardiomyopathy,familial hypertrophic,192600 Creatine phosphokinase,elevated serum,123320 Long QT syndrome 9, 611818 Muscular dystrophy,limb-girdle,type IC,607801 Myopathy,distal,Tateyama type,614321 Rippling muscle disease,606072
CCDC78	92.3	100%	98%	Myopathy, centronuclear, 4, 614807
CFL2	116.8	85%	84%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	123	87%	86%	Myasthenic syndrome, congenital, associated with episodic apnea, 254210
CHCHD10	18.8	42%	35%	?Myopathy,isolated mitochondrial,autosomal dominant,616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2,615911 Spinal muscular atrophy,Jokela type,615048
CHKB	82.4	98%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	114.8	100%	98%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 60893 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	130.2	98%	93%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, slow-channel congenital, 601462
CHRND	144.5	99%	96%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930
CHRNE	123.5	100%	95%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, fast-channel congenital, 608930 Myasthenic syndrome, slow-channel congenital, 601462
CLCN1	136.3	100%	99%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive
CNTN1	158.4	100%	97%	Myopathy, congenital, Compton-North, 612540
COL12A1	136.1	99%	97%	?Ulrich congenital muscular dystrophy 2,616470 Bethlem myopathy 2,616471
COL13A1				Myasthenic syndrome,congenital,19,616720
COL6A1	123.5	100%	98%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 {Ossification of the posterior longitudinal spinal ligaments}, 602475 (2)

COL6A2	132.6	100%	99%	Bethlem myopathy, 158810 Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy, 254090
COL6A3	154.9	100%	99%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090
COLQ	98.1	99%	97%	Endplate acetylcholinesterase deficiency, 603034
CPT2	142.8	96%	93%	CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 Myopathy due to CPT II deficiency, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CRYAB	120.2	97%	95%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
DAG1	180.9	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DES	108.8	100%	99%	?Muscular dystrophy, limb-girdle, type 2R, 615325 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DMD	83.9	99%	94%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DNA2				Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6, 615156
DNAJB6	55.5	89%	75%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNM2	116	96%	93%	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150
DOK7	66	94%	90%	?Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 10, 254300
DPAGT1	112.3	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM2	98.3	100%	100%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	136	100%	100%	Congenital disorder of glycosylation, type Io, 612937

DYNC1H1	166.1	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	124	99%	99%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	80.9	87%	78%	Arthrogryposis,distal,type 5D,615065
EMD	60	97%	84%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	170.5	100%	100%	Glycogen storage disease XIII, 612932
ERBB3	119	99%	98%	Lethal congenital contractural syndrome 2, 607598
EXOSC8	75	85%	72%	Pontocerebellar hypoplasia,type 1C,616081
FAM111B	159.6	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy and pulmonary fibrosis, 615704
FHL1	58.5	96%	75%	Emery-Dreifuss muscular dystrophy 6,X-linked,300696 Myopathy,X-linked,with postural muscle atrophy,300696 Reducing body myopathy,X-linked 1a,severe,infantile or early childhood onset,300717 Reducing body myopathy,X-linked 1b,with late childhood or adult onset,300718 Scapuloperoneal myopathy,X-linked dominant,300695
FKRP	70.4	100%	96%	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
FKTN	137.2	97%	89%	Cardiomyopathy,dilated,1X,611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,4,253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation),type B,4,613152 Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,4,611588
FLNC	149.8	100%	99%	Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
GAA	90.9	100%	99%	Glycogen storage disease II, 232300
GBE1	148.6	100%	93%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	145.8	100%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GNE	150.9	100%	100%	Nonaka myopathy, 605820 Sialuria, 269921
GYG1	128.6	100%	97%	Glycogen storage disease XV, 613507

GYS1	104	100%	97%	Glycogen storage disease 0, muscle, 611556
HSPG2	108	99%	97%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	94.6	100%	87%	Neuronopathy, distal hereditary motor, type VI, 604320
ISCU	123.5	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	107.2	96%	82%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITGA7	115.7	98%	94%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	80.5	100%	94%	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	212.3	100%	100%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome-3, 609622
KLHL41	198.1	100%	98%	Nemaline myopathy 9, 615731
KLHL9	278.3	100%	100%	No OMIM disease
LAMA2	144.2	99%	98%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMP2	76.4	92%	91%	Danon disease, 300257
LARGE	122.9	100%	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	108.4	94%	92%	Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	51.8	89%	81%	Glycogen storage disease XI, 612933
LMNA	72	95%	87%	Cardiomyopathy,dilated,1A,115200 Charcot-Marie-Tooth disease,type 2B1,605588 Emery-Dreifuss muscular dystrophy 2,AD,181350 Emery-Dreifuss muscular dystrophy 3,AR,616516 Heart-hand syndrome,Slovenian type,610140 Hutchinson-Gilford progeria,176670 Lipodystrophy,familial partial,2,151660 Malouf syndrome,212112 Mandibuloacral dysplasia,248370 Muscular dystrophy,congenital,613205 Muscular dystrophy,limb-girdle,type 1B,159001

				Restrictive dermopathy,lethal,275210
LMOD3	160.8	100%	99%	Nemaline myopathy 10,616165
LPIN1	123.7	97%	92%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MATR3	82.2	96%	87%	Myopathy, distal 2, 606070
MEGF10	140.6	100%	99%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MICU1	113.1	94%	89%	Myopathy with extrapyramidal signs, 615673
MSTN	176.6	100%	99%	Muscle hypertrophy, 614160
MTM1	72.4	96%	81%	Myotubular myopathy, X-linked, 310400
MUSK	158.2	100%	100%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
MYF6	81.4	100%	100%	Myopathy, centronuclear, 3, 614408
MYH2	126	100%	99%	Inclusion body myopathy-3, 605637
MYH3	104.2	99%	97%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680
MYH7	106.3	99%	95%	Cardiomyopathy,dilated, 1S,613426 Cardiomyopathy,hypertrophic,1, 192600 Left ventricular noncompaction 5, 613426 Liang distal myopathy,160500 Myopathy,myosin storage,autosomal dominant,608358 Myopathy,myosin storage,autosomal recessive,255160 Scapuloperoneal syndrome,myopathic type,181430
MYOT	144.1	100%	96%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
NEB	130.7	83%	81%	Nemaline myopathy 2, autosomal recessive, 256030

ORAI1	177.8	90%	88%	Immunodeficiency 9,612782 Myopathy,tubular aggregate, 2,615883
PABPN1	70.5	63%	60%	Oculopharyngeal muscular dystrophy, 164300
PFKM	149.2	100%	100%	Glycogen storage disease VII, 232800
PGAM2	138.7	100%	99%	Glycogen storage disease X, 261670
PGK1	35.1	80%	65%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	120.8	100%	99%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHKA1	72.8	94%	90%	? Muscle glycogenosis, 300559
PIP5K1C	96.9	96%	95%	Lethal congenital contractural syndrome 3, 611369
PLEC	87.5	98%	95%	?Epidermolysis bullosa simplex with nail dystrophy,616487 Epidermolysis bullosa simplex, Ogna type,131950 Epidermolysis bullosa simplex with muscular dystrophy,226670 Epidermolysis bullosa simplex with pyloric atresia,612138 Muscular dystrophy,limb-girdle,type 2Q,613723
PNPLA2	104.4	100%	96%	Neutral lipid storage disease with myopathy, 610717
POMGNT1	107	100%	95%	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
POMGNT2	259.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,8),614830
POMK	192.2	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	152.8	100%	97%	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	101.6	97%	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	115.2	100%	97%	Lipodystrophy, congenital generalized, type 4, 613327
PYGM	135.5	100%	99%	McArdle disease, 232600

RAPSN	116.4	94%	93%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency, 608931
RBCK1	89.2	98%	95%	Polyglucosan body myopathy 1 with or without immunodeficiency,615895
RYR1	107.3	95%	91%	{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	209.6	100%	99%	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, acetazolamide-responsive, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SEPN1	95.8	83%	79%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	136.9	100%	100%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	168.1	96%	95%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	81.4	99%	94%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCG	119.5	100%	100%	Muscular dystrophy, limb-girdle, type 2C, 253700
SLC52A2	168	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	104.7	100%	98%	Brown-Vialetto-Van Laere syndrome 1,211530 Fazio-Londe disease,211500
SMCHD1				Fascioscapulohumeral muscular dystrophy 2,digenic,158901
SPEG	78.9	95%	82%	Centronuclear myopathy 5,615959
STIM1	125.1	99%	95%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
TCAP	59.8	100%	100%	Cardiomyopathy, dilated, 1N, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TMEM5	111.8	93%	89%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TNNI2	99.5	100%	99%	Arthrogryposis multiplex congenita, distal, type 2B, 601680

TNNT1	86	97%	93%	Nemaline myopathy 5, Amish type, 605355
TNPO3	137.3	100%	99%	Muscular dystrophy,limb-girdle,type 1F,608423
TPM2	104	100%	97%	Arthrogryposis, distal, type 2B, 601680 Arthrogryposis multiplex congenita, distal, type 1, 108120 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	91.3	89%	88%	CAP myopathy 1, 609284 Myopathy congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRAPPC11	121	96%	95%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRIM32	134.2	100%	100%	Bardet-Biedl syndrome 11, 209900 Muscular dystrophy, limb-girdle, type 2H, 254110
TRPV4	173.3	99%	98%	Brachyolmia type 3, 113500 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Scapuloperoneal spinal muscular atrophy, 181405 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TTC19	90.6	89%	82%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	196	98%	97%	Cardiomyopathy,dilated,1G,604145 Cardiomyopathy,familial hypertrophic,9,613765 Muscular dystrophy,limb-girdle,type 2J,608807 Myopathy,early-onset,with fatal cardiomyopathy,611705 Myopathy,proximal,with early respiratory muscular involvement,603689 Tibial muscular dystrophy,tardive,600334
UBA1	97.2	97%	96%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	147	100%	99%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VIPAS39	135.4	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VRK1	127.4	100%	97%	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

Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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