MUSCLE DISORDERS PANEL DG 2.17 (164 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACADVL	125.2	99.9%	98.7%	VLCAD deficiency, 201475
ACTA1	106.8	99.9%	98.5%	Myopathy, actin, congenital, with cores, 161800 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ACVR1	139.8	100.0%	100.0%	Fibrodysplasia ossificans progressiva, 135100
AGL	141.9	100.0%	99.7%	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGRN	174.0	99.0%	95.9%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	126.7	99.7%	97.1%	Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ATP2A1	157.9	100.0%	100.0%	Brody myopathy, 601003
ATP7A	109.4	99.7%	97.2%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
B3GALNT2	94.8	93.1%	91.1%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B4GAT1	153.5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	189.4	100.0%	99.9%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	166.3	100.0%	99.9%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	125.8	100.0%	99.5%	Centronuclear myopathy 2, 255200
CACNA1S	130.1	100.0%	99.7%	Hypokalemic periodic paralysis, type 1, 170400
CAPN3	103.8	99.4%	97.3%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CASQ1	98.5	100.0%	98.7%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CAV3	233.9	100.0%	100.0%	Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818

				Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072 Cardiomyopathy, familial hypertrophic, 192600
CAVIN1	200.3	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CCDC78	156.1	100.0%	100.0%	?Centronuclear myopathy 4, 614807
CFL2	120.4	100.0%	99.5%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	125.5	97.1%	89.5%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	28.6	67.7%	41.5%	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	96.7	94.5%	93.4%	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	134.8	100.0%	99.8%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	154.8	99.9%	98.3%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 Multiple pterygium syndrome, lethal type, 253290
CHRNE	186.2	100.0%	100.0%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, 4B, fast-channel, 616324
CLCN1	134.2	100.0%	99.9%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CNTN1	128.8	99.9%	99.1%	?Myopathy, congenital, Compton-North, 612540
COL12A1	123.5	99.9%	99.1%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	96.7	100.0%	99.4%	Myasthenic syndrome, congenital, 19, 616720
COL6A1	178.1	100.0%	100.0%	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	195.3	100.0%	99.9%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	163.9	100.0%	99.8%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090

COLQ	106.4	99.9%	98.2%	Myasthenic syndrome, congenital, 5, 603034
CPT2	152.7	98.3%	98.3%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CRYAB	96.3	99.9%	97.9%	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
DAG1	205.3	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538
DES	138.0	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DGUOK	127.0	100.0%	98.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DMD	107.5	99.6%	97.9%	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DNA2	121.9	99.9%	97.8%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJB6	65.0	97.5%	87.0%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNM2	134.2	99.8%	97.7%	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DOK7	156.2	94.4%	93.5%	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence 3, 618389
DPAGT1	93.2	100.0%	100.0%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	134.2	95.5%	87.7%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	95.4	100.0%	99.1%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	218.6	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DYNC1H1	149.0	100.0%	99.8%	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228

DYSF	143.5	100.0%	99.9%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	121.5	100.0%	99.0%	Arthrogryposis, distal, type 5D, 615065
EMD	147.1	99.9%	99.1%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	186.3	100.0%	100.0%	?Glycogen storage disease XIII, 612932
ERBB3	120.0	100.0%	99.3%	?Lethal congenital contractural syndrome 2, 607598
EXOSC8	86.9	98.9%	88.8%	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	155.6	99.9%	99.5%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FHL1	68.2	99.2%	92.8%	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696
FKBP14	77.6	100.0%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	178.0	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	108.0	99.9%	96.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLNC	169.4	100.0%	99.9%	Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Cardiomyopathy, familial hypertrophic, 26, 0
GAA	180.3	100.0%	100.0%	Glycogen storage disease II, 232300
GATM	139.0	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GBE1	152.5	100.0%	99.5%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GFPT1	142.7	99.9%	99.3%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GMPPB	233.1	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNE	115.7	100.0%	99.3%	Sialuria, 269921 Nonaka myopathy, 605820

GRIN1	186.6	100.0%	100.0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GYG1	126.6	100.0%	99.8%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	116.9	100.0%	99.2%	Glycogen storage disease 0, muscle, 611556
HSPG2	132.7	99.5%	99.2%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	117.7	99.9%	98.2%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INPP5K	94.7	100.0%	99.5%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
ISCU	121.3	100.0%	99.9%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	112.0	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITGA7	142.3	99.8%	98.4%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	208.9	100.0%	100.0%	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	164.2	100.0%	100.0%	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KLHL40	144.2	100.0%	100.0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	174.8	100.0%	99.6%	Nemaline myopathy 9, 615731
KLHL9	190.0	100.0%	100.0%	No OMIM Disease ID
LAMA2	131.6	100.0%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMP2	89.8	97.8%	92.3%	Danon disease, 300257
LARGE1	122.8	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LDB3	161.1	96.3%	95.0%	Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493
LDHA	57.3	97.7%	89.0%	Glycogen storage disease XI, 612933
LMNA	118.2	98.3%	93.2%	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210

				Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMOD3	134.2	100.0%	99.9%	Nemaline myopathy 10, 616165
LPIN1	128.5	99.5%	97.0%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MEGF10	129.9	100.0%	99.9%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MICU1	103.4	98.7%	96.2%	Myopathy with extrapyramidal signs, 615673
MLIP	141.9	100.0%	99.4%	No OMIM Disease ID
MSTN	156.1	100.0%	100.0%	Muscle hypertrophy, 614160
MTM1	78.4	98.4%	91.8%	Myotubular myopathy, X-linked, 310400
MUSK	135.2	100.0%	100.0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYF6	181.2	100.0%	100.0%	No OMIM Disease ID
MYH2	113.6	100.0%	99.7%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	100.2	100.0%	99.1%	Contractures, pterygia, and variable skeletal fusions syndrome 1B, 618469 Contractures, pterygia, and variable skeletal fusions syndrome 1A, 178110 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYH7	101.7	99.7%	97.2%	Myopathy, myosin storage, autosomal recessive, 255160 Left ventricular noncompaction 5, 613426 Laing distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Cardiomyopathy, hypertrophic, 1, 192600
MYOT	142.2	100.0%	99.1%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYPN	128.9	99.9%	99.0%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
NEB	101.5	83.0%	82.5%	Nemaline myopathy 2, autosomal recessive, 256030
NEFH	121.3	99.5%	98.0%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
OPA1	121.4	99.7%	97.5%	Behr syndrome, 210000 Optic atrophy 1, 165500

				Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
ORAI1	226.3	100.0%	99.0%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
PABPN1	82.7	82.5%	63.4%	Oculopharyngeal muscular dystrophy, 164300
PFKM	119.4	99.9%	99.3%	Glycogen storage disease VII, 232800
PGAM2	180.7	100.0%	100.0%	Glycogen storage disease X, 261670
PGK1	47.0	92.1%	78.7%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	134.8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PHKA1	90.2	97.8%	91.7%	Muscle glycogenosis, 300559
PIEZO2	106.9	99.9%	99.4%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIP5K1C	151.5	99.9%	98.4%	Lethal congenital contractural syndrome 3, 611369
PLEC	165.0	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950
PNPLA2	159.9	100.0%	99.9%	Neutral lipid storage disease with myopathy, 610717
POMGNT1	123.6	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	225.4	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	144.2	100.0%	100.0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	137.5	99.6%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 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	109.7	100.0%	99.1%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
PRPS1	113.2	100.0%	99.9%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500

				Arts syndrome, 301835 Gout, PRPS-related, 300661
PYGM	130.4	100.0%	100.0%	McArdle disease, 232600
RAPSN	162.2	99.9%	99.2%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	118.4	100.0%	99.4%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RRM2B	142.6	100.0%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RYR1	128.2	99.3%	96.8%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
SCN4A	182.1	99.9%	99.4%	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SELENON	143.9	85.3%	84.0%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	170.3	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	135.1	99.8%	97.9%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	78.2	100.0%	98.2%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	117.7	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SLC25A4	141.9	100.0%	99.9%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC52A2	213.2	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	128.9	100.0%	99.9%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SMCHD1	95.2	99.6%	97.4%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 Bosma arhinia microphthalmia syndrome, 603457
SPEG	145.0	99.5%	98.1%	Centronuclear myopathy 5, 615959
SRPK3	142.2	99.9%	98.8%	No OMIM Disease ID
STIM1	129.2	99.8%	97.1%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070

TANGO2	139.6	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TCAP	113.4	100.0%	100.0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TK2	111.5	100.0%	99.8%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM5	167.7	99.8%	96.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TNNI2	169.0	100.0%	100.0%	Arthrogryposis, distal, type 2B1, 601680
TNNT1	113.8	100.0%	99.4%	Nemaline myopathy 5, Amish type, 605355
TNPO3	116.9	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TPM2	114.5	100.0%	99.8%	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
TPM3	76.2	89.4%	88.3%	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TRAPPC11	124.3	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRIM32	132.8	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIP4	105.3	100.0%	98.9%	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRPV4	150.4	100.0%	100.0%	Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Scapuloperoneal spinal muscular atrophy, 181405 Metatropic dysplasia, 156530 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Brachyolmia type 3, 113500 ?Avascular necrosis of femoral head, primary, 2, 617383
TTC19	84.9	98.8%	86.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	165.0	98.6%	98.1%	Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, familial hypertrophic, 9, 613765

				Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807
TWNK	170.3	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
UBA1	139.8	99.7%	98.4%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	103.9	100.0%	99.4%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VIPAS39	114.4	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	97.5	99.4%	95.0%	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	126.7	99.9%	98.3%	Pontocerebellar hypoplasia type 1A, 607596
XK	88.8	99.9%	99.5%	McLeod syndrome with or without chronic granulomatous disease, 300842
YARS2	188.2	100.0%	99.4%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZC4H2	74.3	99.6%	95.6%	Wieacker-Wolff syndrome, 314580

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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors