MUSCLE DISORDERS GENE PANEL DG 2.15 (159 genes)

Releasedate: 31-01-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACADVL	118.8	98.7	95.1	VLCAD deficiency, 201475
ACTA1	99.7	99.2	95.3	?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	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100
AGL	146.7	99.7	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	114.8	95.2	89.3	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	142.2	99.5	95.9	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ATP2A1	155.9	100	100	Brody myopathy, 601003
ATP7A	133.2	99.7	97.8	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
B3GALNT2	115	92.4	89.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B4GAT1	120.4	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	136.5	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	158.6	100	99.9	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	100.2	99.1	95.3	Centronuclear myopathy 2, 255200
CACNA1S	135.5	100	99.7	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CAPN3	111.4	99	96.7	Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600

CASQ1	122	100	99.6	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CAV3	304.7	100	100	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072
CAVIN1	137	99.9	99.3	Lipodystrophy, congenital generalized, type 4, 613327
CCDC78	114.9	100	100	?Centronuclear myopathy 4, 614807
CFL2	117.3	94.3	86.7	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	130.3	89.3	86.8	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	20	43	35.2	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	121.8	94.7	94.6	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNB1	131.8	98.8	96.7	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	150.5	100	99	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322
CHRNE	127.7	99.3	95.8	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CLCN1	137.4	100	99.5	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CNTN1	151.7	99.8	98.3	?Myopathy, congenital, Compton-North, 612540
COL12A1	137.5	99.5	97.5	?Ullrich congenital muscular dystrophy 2, 616470 Bethlem myopathy 2, 616471
COL13A1	85.4	99.8	97.1	Myasthenic syndrome, congenital, 19, 616720
COL6A1	137.2	99.5	97.8	Bethlem myopathy 1, 158810

				Ullrich congenital muscular dystrophy 1, 254090
COL6A2	165.3	99.3	98.4	?Myosclerosis, congenital, 255600 Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	174.7	100	99.9	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COLQ	113.2	99.8	98.1	Myasthenic syndrome, congenital, 5, 603034
CPT2	162.8	97.2	95.4	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CRYAB	125.7	99.9	98.7	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
DAG1	220.8	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DES	120.8	99.9	98.1	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DGUOK	119.2	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DMD	112.4	99.4	97.4	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DNA2	123.6	99.8	96.9	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJB6	59.8	91.7	79.3	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNM2	127.4	97.5	94.4	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482

				Lethal congenital contracture syndrome 5, 615368
DOK7	105.7	93.3	92.5	?Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 10, 254300
DPAGT1	110.7	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	131.1	91.7	86.7	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.1	100	99.4	Congenital disorder of glycosylation, type Iu, 615042
DPM3	183.9	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DYNC1H1	179.8	100	99.6	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYSF	133.1	100	99.8	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	100.7	88.8	83.1	Arthrogryposis, distal, type 5D, 615065
EMD	100.3	99.8	97.2	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	179.3	100	100	?Glycogen storage disease XIII, 612932
ERBB3	139.2	100	99.9	?Lethal congenital contractural syndrome 2, 607598 {?Erythroleukemia, familial, susceptibility to}, 133180
EXOSC8	80.1	91.8	76.9	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	152.9	100	99.8	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FHL1	87.2	98.8	93	?Uruguay faciocardiomusculoskeletal syndrome, 300280 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
FKBP14	74.3	100	99.4	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	94.5	100	99.7	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	120	99.2	94.2	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	165	100	99.7	Cardiomyopathy, familial hypertrophic, 26, 0 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
GAA	128.5	100	99.9	Glycogen storage disease II, 232300
GBE1	145.5	99.6	97.2	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	144.4	99.9	97.6	Myasthenia, congenital, 12, with tubular aggregates, 610542
GMPPB	228.5	100	100	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	153.7	100	99.8	Nonaka myopathy, 605820 Sialuria, 269921
GRIN1	150.7	100	99.5	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GYG1	157.8	100	99.6	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	110.8	100	98.5	Glycogen storage disease 0, muscle, 611556
HSPG2	121.3	99.4	98.2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	107.8	99.3	96	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
INPP5K	108.3	100	99.6	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
ISCU	111.2	100	99.7	Myopathy with lactic acidosis, hereditary, 255125
ISPD	104.4	95.2	84.8	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITGA7	129.6	99.6	97.6	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	107.1	99.8	96.8	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	229.3	100	100	Andersen syndrome, 170390

				Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KLHL40	157.9	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	203.5	100	99.6	Nemaline myopathy 9, 615731
KLHL9	260.8	100	100	No OMIM phenotype Myopathy, distal, early-onset (Cirak (2010) Brain 133, 2123)
LAMA2	143.5	99.9	99.5	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMP2	106.1	92.7	91.2	Danon disease, 300257
LARGE1	143	100	99.6	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	127.3	95.5	93.7	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	59.8	94.1	87.1	Glycogen storage disease XI, 612933
LMNA	89.2	97.9	91.3	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMOD3	141.5	99.9	98.5	Nemaline myopathy 10, 616165
LPIN1	134.6	99.8	97.8	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MB	153.8	100	100	No OMIM phenotype
MEGF10	154.3	100	99.8	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MICU1	134.2	96	88.8	Myopathy with extrapyramidal signs, 615673
MSTN	161.5	100	99.5	Muscle hypertrophy, 614160

MTM1	93.7	99.2	93	Myotubular myopathy, X-linked, 310400
MUSK	159.4	100	99.9	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYF6	121.6	100	100	Centronuclear myopathy 3, 614408
MYH2	129.4	99.9	98.8	Proximal myopathy and ophthalmoplegia, 605637
MYH3	110.4	99.9	98.6	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
MYH7	111.4	99.4	96.8	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYOT	139.4	99.3	95.5	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
NEB	124	82.9	81.9	Nemaline myopathy 2, autosomal recessive, 256030
OPA1	122.5	99.1	94.1	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	237.3	93.8	89.8	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
PABPN1	66.6	62.4	60.5	Oculopharyngeal muscular dystrophy, 164300
PFKM	150.4	100	99.8	Glycogen storage disease VII, 232800
PGAM2	170.9	100	99.9	Glycogen storage disease X, 261670
PGK1	54.5	93.3	81.6	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	133.6	100	99.9	Congenital disorder of glycosylation, type It, 614921
PHKA1	106.7	98.9	95.3	Muscle glycogenosis, 300559
PIP5K1C	107.6	96.3	95.1	Lethal congenital contractural syndrome 3, 611369
PLEC	114.1	99.7	98.7	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670

				Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PNPLA2	113.2	99.7	97.4	Neutral lipid storage disease with myopathy, 610717
POMGNT1	127.6	99.7	97.1	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 Retinitis pigmentosa 76, 617123
POMGNT2	259.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	205.1	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	155.7	99.7	98.1	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	111.1	98.9	97.5	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
PRPS1	149.5	100	100	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PYGM	127.1	100	99.9	McArdle disease, 232600
RAPSN	140.5	99.6	96.3	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	104.1	99.2	94.9	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RRM2B	128.6	99.7	97.5	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RYR1	120.7	96.8	93.7	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000

				{Malignant hyperthermia susceptibility 1}, 145600
SCN4A	214	99.9	99.5	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SELENON	111.7	85.2	83.3	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	147.3	100	99.7	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	154.2	96.6	94.2	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94.8	100	99.4	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	138.7	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SLC25A4	134.1	100	100	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC52A2	177.6	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.6	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SMCHD1	91.3	98.1	92.3	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SPEG	100.9	93.3	86.5	Centronuclear myopathy 5, 615959
STIM1	145.3	100	99.2	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TCAP	89	100	99.2	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TK2	105.7	93.4	89.4	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM5	120.5	96.8	92.9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041

TNNI2	121.2	100	99.6	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNT1	86.9	96.3	94	Nemaline myopathy 5, Amish type, 605355
TNPO3	139.6	100	99.7	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TPM2	109.1	100	99.6	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	98.9	89.4	89.1	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRAPPC11	126.2	99.4	96.4	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRIM32	141.2	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIP4	113.5	100	98.8	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRPV4	172.4	99.5	98.7	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TTC19	92.1	80.6	72.5	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	187.8	98.2	97.2	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334
TWNK	178.8	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138

				Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
UBA1	162	99.8	98.9	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	144.8	99.9	99.5	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VIPAS39	144.6	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	42.3	95.2	81.3	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	124.8	97.5	94.2	Pontocerebellar hypoplasia type 1A, 607596
XK	96.8	99.9	99.1	McLeod syndrome with or without chronic granulomatous disease, 300842
YARS2	173.2	99.8	98.9	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZC4H2	78.6	99.8	98.1	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 31st 2018.
This list is accurate for panel version DG 2.15
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors