MUSCLE DISORDERS GENE PANEL DG 2.18 (167 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACADVL	99,40%	97,30%	100%	100%	VLCAD deficiency, 201475
ACTA1	99,60%	92,30%	100%	100%	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	100%	100%	100%	100%	Fibrodysplasia ossificans progressiva, 135100
AGL	100%	99,40%	100%	100%	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGRN	96,90%	92,60%	100%	99,90%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	99,50%	97,30%	100%	100%	Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ATP2A1	100%	100%	100%	100%	Brody myopathy, 601003
ATP7A	99,70%	97,50%	100%	100%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
B3GALNT2	93,80%	89,40%	92,50%	92,50%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B4GAT1	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	100%	100%	100%	100%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	100%	99,70%	100%	100%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	99,60%	95,70%	100%	100%	Centronuclear myopathy 2, 255200
CACNA1S	100%	99,90%	100%	100%	Hypokalemic periodic paralysis, type 1, 170400
CAPN3	100%	99,30%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CASQ1	100%	99,50%	100%	100%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CAV3	100%	100%	100%	100%	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	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
CCDC78	100%	100%	100%	100%	?Centronuclear myopathy 4, 614807
CFL2	100%	99,60%	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	93,50%	85,70%	100%	100%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	59,10%	43,90%	100%	100%	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHKB	100%	99,70%	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	94,70%	94,00%	100%	100%	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	100%	99,40%	100%	100%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	99,70%	97,90%	100%	100%	?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	100%	100%	100%	100%	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	100%	99,20%	100%	100%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CNTN1	99,90%	98,90%	100%	100%	?Myopathy, congenital, Compton-North, 612540
COL12A1	100%	99,40%	100%	100%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100%	99,80%	100%	100%	Myasthenic syndrome, congenital, 19, 616720
COL6A1	100%	99,40%	100%	100%	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	100%	99,80%	100%	100%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100%	99,80%	100%	100%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090

COLQ	100%	99,20%	100%	100%	Myasthenic syndrome, congenital, 5, 603034
CPT2	98,20%	97,80%	100%	100%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CRYAB	100%	99,20%	100%	100%	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
DAG1	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538
DES	100%	99,70%	100%	100%	?Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DGUOK	100%	99,40%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DMD	99,60%	98,60%	100%	100%	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DNA2	99,80%	98,30%	100%	100%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJB6	96,50%	88,50%	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNM2	98,10%	94,50%	100%	100%	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	94,10%	91,30%	100%	100%	Myasthenic syndrome, congenital, 10, 254300 Fetal akinesia deformation sequence 3, 618389
DPAGT1	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	98,20%	91,30%	99,70%	97,10%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	98,70%	100%	100%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DYNC1H1	99,90%	99,40%	100%	100%	Mental retardation, autosomal dominant 13, 614563 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600

DYSF	100%	99,90%	100%	100%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	95,40%	90,00%	100%	100%	Arthrogryposis, distal, type 5D, 615065
EMD	99,90%	98,40%	100%	99,10%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	100%	99,90%	100%	100%	?Glycogen storage disease XIII, 612932
ERBB3	100%	99,80%	100%	100%	?Lethal congenital contractural syndrome 2, 607598
EXOSC8	97,90%	91,20%	100%	100%	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	100%	99,90%	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FHL1	99,70%	95,80%	100%	100%	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	100%	99,90%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100%	100%	100%	99,90%	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	99,70%	97,00%	100%	100%	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	100%	99,60%	100%	100%	Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Cardiomyopathy, familial hypertrophic, 26, 0
GAA	100%	99,90%	100%	100%	Glycogen storage disease II, 232300
GATM	100%	100%	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
GBE1	100%	99,60%	100%	100%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GFPT1	100%	99,40%	100%	100%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GMPPB	100%	100%	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	100%	99,70%	100%	100%	Sialuria, 269921 Nonaka myopathy, 605820

GRIN1	100%	100%	100%	100%	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	99,90%	99,20%	100%	100%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100%	98,60%	100%	100%	Glycogen storage disease 0, muscle, 611556
HSPG2	99,20%	97,70%	100%	99,90%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	98,80%	95,10%	100%	100%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INPP5K	100%	100%	100%	100%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
ISCU	100%	100%	100%	100%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	98,50%	94,80%	100%	99,40%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITGA7	99,60%	98,00%	100%	100%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	99,80%	95,80%	100%	100%	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	100%	100%	100%	100%	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KIF21A	99,90%	99,30%	100%	100%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KLHL40	100%	100%	100%	100%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	99,90%	100%	100%	Nemaline myopathy 9, 615731
KLHL9	100%	100%	100%	100%	No OMIM disease ID
LAMA2	100%	99,60%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMP2	99,20%	95,60%	100%	100%	Danon disease, 300257
LARGE1	100%	99,60%	100%	100%	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	95,40%	94,70%	100%	100%	Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493
LDHA	95,00%	91,70%	100%	100%	Glycogen storage disease XI, 612933
LMNA	97,40%	91,90%	100%	100%	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	100%	99,70%	100%	100%	Nemaline myopathy 10, 616165
LPIN1	99,60%	97,30%	100%	100%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MEGF10	100%	100%	100%	100%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MICU1	98,90%	95,20%	100%	100%	Myopathy with extrapyramidal signs, 615673
MLIP	99,90%	99,00%	100%	100%	No OMIM disease ID
MSTN	100%	100%	100%	100%	Muscle hypertrophy, 614160
MTM1	99,00%	93,30%	100%	100%	Myotubular myopathy, X-linked, 310400
MUSK	100%	99,90%	100%	100%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYF6	100%	100%	100%	100%	No OMIM disease ID
MYH2	99,90%	99,40%	100%	100%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	99,90%	99,00%	100%	100%	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110
MYH7	99,60%	97,30%	100%	100%	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	100%	99,60%	100%	100%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYPN	100%	99,70%	100%	100%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
NEB	83,00%	82,60%	99,90%	99,90%	Nemaline myopathy 2, autosomal recessive, 256030

NEFH	93,40%	84,50%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
OPA1	99,70%	97,60%	100%	100%	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
ORAI1	95,80%	92,80%	97,20%	92,40%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
PABPN1	66,30%	56,90%	100%	99,10%	Oculopharyngeal muscular dystrophy, 164300
PFKM	100%	99,50%	100%	100%	Glycogen storage disease VII, 232800
PGAM2	100%	100%	100%	100%	Glycogen storage disease X, 261670
PGK1	92,80%	79,30%	100%	100%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	100%	100%	100%	100%	Congenital disorder of glycosylation, type It, 614921
PHKA1	99,20%	95,30%	100%	99,90%	Muscle glycogenosis, 300559
TANGO2	100%	99,30%	100%	100%	metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
PIEZO2	100%	99,50%	100%	100%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIP5K1C	98,00%	95,80%	99,90%	99,80%	Lethal congenital contractural syndrome 3, 611369
PLEC	100%	99,80%	100%	100%	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	99,70%	96,10%	100%	100%	Neutral lipid storage disease with myopathy, 610717
POMGNT1	100%	99,90%	100%	100%	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	100%	100%	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	100%	100%	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	99,30%	97,50%	100%	100%	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	99,40%	96,40%	100%	100%	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	100%	100%	100%	100%	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	100%	99,90%	100%	100%	McArdle disease, 232600
RAPSN	100%	99,70%	100%	100%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	99,90%	98,20%	100%	100%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RRM2B	100%	99,70%	100%	100%	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	96,90%	93,90%	99,40%	99,00%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
SCN4A	100%	99,60%	100%	100%	Paramyotonia congenita, 168300 Hyperkalemic periodic paralysis, type 2, 170500 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SELENON	84,50%	84,00%	87,70%	85,10%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	100%	99,90%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	97,70%	96,50%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100%	98,90%	100%	100%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100%	99,20%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SLC25A4	100%	100%	100%	100%	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	100%	100%	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500

SMCHD1	99,50%	96,30%	100%	100%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 Bosma arhinia microphthalmia syndrome, 603457
SPEG	96,40%	89,50%	99,70%	99,70%	Centronuclear myopathy 5, 615959
SRPK3	98,70%	96,10%	100%	100%	No OMIM disease ID
STIM1	99,80%	98,00%	100%	100%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
SYT2	99,90%	99,00%	100%	100%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
TCAP	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TK2	99,20%	96,30%	100%	100%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM5	99,50%	96,80%	100%	99,90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TNNI2	100%	99,70%	100%	100%	Arthrogryposis, distal, type 2B1, 601680
TNNT1	99,90%	97,60%	100%	100%	Nemaline myopathy 5, Amish type, 605355
TNPO3	100%	99,90%	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TPM2	100%	100%	84,00%	83,70%	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
TPM3	89,20%	87,20%	100%	100%	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TRAPPC11	100%	99,20%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRIM32	100%	100%	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIP4	100%	99,10%	100%	100%	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRPV4	100%	99,90%	100%	100%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 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	81,50%	73,80%	100%	99,20%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	98,60%	98,10%	100%	100%	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
TUBB3	98,30%	96,90%	100%	100%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
UBA1	99,40%	98,20%	99,80%	99,00%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	100%	99,20%	100%	100%	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	100%	100%	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	99,00%	94,60%	100%	98,60%	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	99,70%	98,50%	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
XK	99,80%	98,10%	100%	100%	McLeod syndrome with or without chronic granulomatous disease, 300842
YARS2	100%	99,80%	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZC4H2	100%	99,00%	100%	100%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors