MUSCLE DISORDERS GENE PANEL DG 3.4.0 (180 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACADVL	100,0%	100,0%	VLCAD deficiency, 201475
ACTA1	100,0%	100,0%	?Myopathy, scapulohumeroperoneal, 616852 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	100,0%	100,0%	Fibrodysplasia ossificans progressiva, 135100
AGL	100,0%	100,0%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	100,0%	100,0%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260
ASCC1	87,1%	87,1%	Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ATP2A1	100,0%	100,0%	Brody myopathy, 601003
ATP7A	100,0%	100,0%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
B3GALNT2	92,5%	92,5%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B4GAT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	100,0%	100,0%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	100,0%	100,0%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	100,0%	100,0%	Centronuclear myopathy 2, 255200
CACNA1S	100,0%	100,0%	Hypokalemic periodic paralysis, type 1, 170400
CAPN3	97,9%	97,9%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129

CASQ1	100,0%	100,0%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CAV3	100,0%	100,0%	Myopathy, distal, Tateyama type, 614321 Creatine phosphokinase, elevated serum, 123320 Cardiomyopathy, familial hypertrophic, 192600 Rippling muscle disease 2, 606072 Long QT syndrome 9, 611818
CAVIN1	100,0%	100,0%	Lipodystrophy, congenital generalized, type 4, 613327
CCDC78	100,0%	100,0%	?Centronuclear myopathy 4, 614807
CFL2	100,0%	100,0%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	100,0%	100,0%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	100,0%	100,0%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHKB	100,0%	100,0%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	100,0%	100,0%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	100,0%	100,0%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	100,0%	100,0%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321
CHRNE	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
CHRNG	100,0%	100,0%	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CLCN1	100,0%	100,0%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive,
CNTN1	100,0%	100,0%	?Myopathy, congenital, Compton-North, 612540
COL12A1	100,0%	100,0%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100,0%	100,0%	Myasthenic syndrome, congenital, 19, 616720
COL6A1	100,0%	100,0%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090

COL6A2	100,0%	100,0%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100,0%	100,0%	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810
COLQ	100,0%	100,0%	Myasthenic syndrome, congenital, 5, 603034
CPT2	100,0%	100,0%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CRPPA	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRYAB	100,0%	100,0%	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Cardiomyopathy, dilated, 1II, 615184
DAG1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS2	100,0%	100,0%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DES	100,0%	100,0%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DGUOK	100,0%	100,0%	Portal hypertension, noncirrhotic, 1, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DMD	100,0%	100,0%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DNA2	100,0%	100,0%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJB6	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNM2	100,0%	100,0%	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	100,0%	100,0%	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300

DPAGT1	100,0%	100,0%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPM1	99,8%	97,8%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100,0%	100,0%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100,0%	100,0%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DYNC1H1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Intellectual developmental disorder, autosomal dominant 13, 614563
DYSF	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Miyoshi muscular dystrophy 1, 254130 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	100,0%	100,0%	Arthrogryposis, distal, type 5D, 615065
EMD	100,0%	100,0%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	100,0%	100,0%	Glycogen storage disease XIII, 612932
ERBB3	100,0%	100,0%	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
EXOSC8	100,0%	100,0%	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	100,0%	100,0%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FHL1	100,0%	100,0%	Myopathy, X-linked, with postural muscle atrophy, 300696 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717
FKBP14	100,0%	100,0%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 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	100,0%	100,0%	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
GAA	100,0%	100,0%	Glycogen storage disease II, 232300

GATM	100,0%	100,0%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBE1	100,0%	100,0%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	100,0%	100,0%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GMPPB	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GNE	100,0%	100,0%	Sialuria, 269921 Nonaka myopathy, 605820
GRIN1	100,0%	100,0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Developmental and epileptic encephalopathy 101, 619814 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GYG1	100,0%	100,0%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100,0%	100,0%	Glycogen storage disease 0, muscle, 611556
HSPG2	100,0%	100,0%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INPP5K	100,0%	100,0%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
ISCU	100,0%	100,0%	Myopathy with lactic acidosis, hereditary, 255125
ITGA7	100,0%	100,0%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	100,0%	100,0%	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	100,0%	100,0%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KIF21A	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 3B, 135700 Fibrosis of extraocular muscles, congenital, 1, 135700
KLHL40	100,0%	100,0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100,0%	100,0%	Nemaline myopathy 9, 615731
KLHL9	100,0%	100,0%	No OMIM Disease ID
LAMA2	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMP2	100,0%	100,0%	Danon disease, 300257
LARGE1	100,0%	100,0%	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	100,0%	100,0%	Left ventricular noncompaction 3, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LDHA	100,0%	100,0%	Glycogen storage disease XI, 612933
LMNA	100,0%	100,0%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Restrictive dermopathy 2, 619793 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LMOD3	100,0%	100,0%	Nemaline myopathy 10, 616165
LPIN1	100,0%	100,0%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MAP3K20	100,0%	100,0%	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MEGF10	100,0%	100,0%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MICU1	100,0%	100,0%	Myopathy with extrapyramidal signs, 615673
MLIP	100,0%	100,0%	No OMIM Disease ID
MSTN	100,0%	100,0%	?Muscle hypertrophy, 614160
MTM1	100,0%	100,0%	Myopathy, centronuclear, X-linked, 310400
MUSK	100,0%	100,0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYH2	100,0%	100,0%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	100,0%	100,0%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYH7	100,0%	100,0%	Laing distal myopathy, 160500 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430

			Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160
MYOT	100,0%	100,0%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYPN	100,0%	100,0%	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336
NEB	99,9%	99,9%	Nemaline myopathy 2, autosomal recessive, 256030 Arthrogryposis multiplex congenita 6, 619334
NEFH	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
OPA1	100,0%	100,0%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
ORAI1	100,0%	100,0%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
PABPN1	100,0%	100,0%	Oculopharyngeal muscular dystrophy, 164300
PFKM	100,0%	100,0%	Glycogen storage disease VII, 232800
PGAM2	100,0%	100,0%	Glycogen storage disease X, 261670
PGK1	100,0%	100,0%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94,2%	94,2%	Congenital disorder of glycosylation, type It, 614921
PHKA1	100,0%	99,9%	Muscle glycogenosis, 300559
PHOX2A	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 2, 602078
PIEZO2	100,0%	100,0%	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PIP5K1C	100,0%	100,0%	Lethal congenital contractural syndrome 3, 611369
PLEC	100,0%	100,0%	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 Epidermolysis bullosa simplex 5A, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PNPLA2	100,0%	100,0%	Neutral lipid storage disease with myopathy, 610717
POLG	100,0%	100,0%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700

			Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	100,0%	100,0%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POMGNT1	100,0%	100,0%	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	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	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	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150
PREPL	100,0%	100,0%	Myasthenic syndrome, congenital, 22, 616224
PRPS1	100,0%	100,0%	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PTRH2	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PYGM	100,0%	100,0%	McArdle disease, 232600
RAPSN	100,0%	100,0%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	100,0%	100,0%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RRM2B	100,0%	100,0%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RXYLT1	100,0%	100,0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	100,0%	99,9%	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000

			King-Denborough syndrome, 619542 Minicore myopathy with external ophthalmoplegia, 255320
SCN4A	100,0%	100,0%	Paramyotonia congenita, 168300 Hypokalemic periodic paralysis, type 2, 613345 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hyperkalemic periodic paralysis, type 2, 170500
SELENON	93,0%	91,5%	Myopathy, congenital, with fiber-type disproportion, 255310 Muscular dystrophy, rigid spine, 1, 602771
SGCA	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100,0%	100,0%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SIL1	100,0%	100,0%	Marinesco-Sjogren syndrome, 248800
SLC25A4	100,0%	100,0%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184
SLC52A2	100,0%	100,0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100,0%	100,0%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SMCHD1	100,0%	100,0%	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMDT1	100,0%	100,0%	No OMIM Disease ID
SPEG	100,0%	99,8%	Centronuclear myopathy 5, 615959
SRPK3	100,0%	100,0%	No OMIM Disease ID
STIM1	100,0%	100,0%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
SYT2	100,0%	100,0%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
TANGO2	100,0%	100,0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TCAP	100,0%	100,0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TK2	100,0%	100,0%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TNNI2	100,0%	100,0%	Arthrogryposis, distal, type 2B1, 601680

TNNT1	100,0%	100,0%	Nemaline myopathy 5, Amish type, 605355
TNPO3	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6B	100,0%	100,0%	Global developmental delay with speech and behavioral abnormalities, 619243
TPM2	100,0%	100,0%	Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	100,0%	100,0%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRAPPC11	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRIM32	100,0%	100,0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIP4	100,0%	100,0%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRPV4	100,0%	100,0%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Digital arthropathy-brachydactyly, familial, 606835 SED, Maroteaux type, 184095 Metatropic dysplasia, 156530 Scapuloperoneal spinal muscular atrophy, 181405 Hereditary motor and sensory neuropathy, type IIc, 606071 ?Avascular necrosis of femoral head, primary, 2, 617383 Neuronopathy, distal hereditary motor, type VIII, 600175 Parastremmatic dwarfism, 168400 Brachyolmia type 3, 113500
TTC19	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, familial hypertrophic, 9, 613765 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, dilated, 1G, 604145 Myopathy, myofibrillar, 9, with early respiratory failure, 603689
TUBB3	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	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	100,0%	99,8%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UNC45B	100,0%	100,0%	?Cataract 43, 616279 Myofibrillar myopathy 11, 619178
VCP	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VIPAS39	100,0%	100,0%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	100,0%	100,0%	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	100,0%	100,0%	Pontocerebellar hypoplasia type 1A, 607596
VWA1	100,0%	100,0%	Neuropathy, hereditary motor, with myopathic features, 619216
XK	100,0%	100,0%	McLeod syndrome with or without chronic granulomatous disease, 300842
YARS2	100,0%	100,0%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZC4H2	100,0%	100,0%	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.
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.
TWIST is the chemistry used for 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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors