MUSCLE DISORDERS GENE PANEL DG 3.2.0 (177 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ACADVL	99,7	96,6	100	100	VLCAD deficiency, 201475
ACTA1	98,2	89,5	100	100	?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	99,9	100	100	Fibrodysplasia ossificans progressiva, 135100
AGL	99,8	99,5	100	100	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	97,6	92,6	100	99,9	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	99,2	96,9	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260
ATP2A1	100	100	100	100	Brody myopathy, 601003
ATP7A	98,7	96	100	99,9	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
B3GALNT2	94,3	89,8	92,5	92,5	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	99,7	100	100	Cardiomyopathy, dilated, 1HYPOGONADOTROPIC HYPOGONADISM, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	99,9	99,1	100	100	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	99,7	96	100	100	Centronuclear myopathy 2, 255200
CACNA1S	100	99,8	100	100	Hypokalemic periodic paralysis, type 1, 170400
CAPN3	97,7	96,3	97,9	97,9	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129

CASQ1	99,8	96,5	100	100	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CAV3	100	100	100	100	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	100	100	100	Lipodystrophy, congenital generalized, type 4, 613327
CCDC78	100	100	100	100	?Centronuclear myopathy 4, 614807
CFL2	99,5	99	100	99,9	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	93,1	85,1	100	99,9	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	57,8	42	100	100	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHKB	100	99,6	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	100	99,6	100	100	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	100	99,5	100	100	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	99,4	97,4	100	100	?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	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
CHRNG	100	100	100	100	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CLCN1	99,9	98,8	100	100	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive,
CNTN1	99,7	98,6	100	100	?Myopathy, congenital, Compton-North, 612540
COL12A1	99,9	99,3	100	100	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	93,9	93,5	100	100	Myasthenic syndrome, congenital, 19, 616720

COL6A1	100	99,7	100	100	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	100	99,8	100	100	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100	99,7	100	100	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810
COLQ	99,8	97,1	100	100	Myasthenic syndrome, congenital, 5, 603034
CPT2	98,2	97,4	100	100	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CRPPA	98,4	94,7	100	99,8	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRYAB	100	98,2	100	100	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	99,9	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS2	94,8	93,8	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DES	100	99,6	100	100	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DGUOK	99,9	98,8	100	100	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	99,5	98,1	100	99,9	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DNA2	99,6	96,9	100	100	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJB6	95,9	84,6	100	100	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNM2	98,6	93,9	100	100	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	94,9	92	100	100	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DPAGT1	100	99,8	100	100	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPM1	97,4	90,9	98,6	94,6	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100	97,7	100	100	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100	100	100	100	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DYNC1H1	99,9	99,3	100	100	Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563
DYSF	100	99,8	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Miyoshi muscular dystrophy 1, 254130 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	95,9	91,8	100	100	Arthrogryposis, distal, type 5D, 615065
EMD	99,8	97,9	100	99,8	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	100	100	100	100	?Glycogen storage disease XIII, 612932
ERBB3	100	99,3	100	100	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
EXOSC8	98,7	90	100	100	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	99,9	99,7	100	100	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FHL1	99,4	93,8	100	100	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	99,8	98,7	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100	100	100	100	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	99,8	95,2	100	100	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	99,4	100	100	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
GAA	100	99,9	100	100	Glycogen storage disease II, 232300
GATM	100	100	100	100	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBE1	99,9	99,7	100	100	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	99,9	99,4	100	100	Myasthenia, congenital, 12, with tubular aggregates, 610542
GMPPB	100	100	100	100	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	99,5	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GRIN1	100	99,9	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	99,6	97,4	100	100	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100	98	100	100	Glycogen storage disease 0, muscle, 611556
HSPG2	99,2	97,5	100	99,8	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	99,3	96,9	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INPP5K	100	99,7	100	100	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
ISCU	100	100	100	100	Myopathy with lactic acidosis, hereditary, 255125
ITGA7	99,7	97,9	100	100	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	99,9	96,7	100	100	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	100	100	100	100	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622

KIF21A	99,7	98,9	100	100	Fibrosis of extraocular muscles, congenital, 3B, 135700 Fibrosis of extraocular muscles, congenital, 1, 135700
KLHL40	100	100	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	99,9	99,4	100	100	Nemaline myopathy 9, 615731
KLHL9	100	99,9	100	100	No OMIM disease ID
LAMA2	99,9	99,1	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMP2	99,3	96	100	99,7	Danon disease, 300257
LARGE1	100	99,7	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	95,4	94,8	100	100	Left ventricular noncompaction 3, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LDHA	94,4	89,3	100	100	Glycogen storage disease XI, 612933
LMNA	96,1	90,6	100	100	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Restrictive dermopathy, lethal, 275210 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 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	99,8	99	100	100	Nemaline myopathy 10, 616165
LPIN1	99,4	97,2	100	100	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MAP3K20	99,9	99,2	100	99,9	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MEGF10	100	99,9	100	100	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MICU1	97,3	92,2	100	100	Myopathy with extrapyramidal signs, 615673
MLIP	99,9	98,8	100	100	No OMIM disease ID
MSTN	100	99,9	100	100	?Muscle hypertrophy, 614160

MTM1	98,7	92	100	99,7	Myotubular myopathy, X-linked, 310400
MUSK	100	99,9	100	100	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYH2	99,9	99,1	100	100	Proximal myopathy and ophthalmoplegia, 605637
MYH3	99,9	98,4	100	100	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	99,1	96,7	100	100	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	99,2	100	100	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYPN	100	99,5	100	100	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336
NEB	82,9	82,5	99,9	99,8	Nemaline myopathy 2, autosomal recessive, 256030 Arthrogryposis multiplex congenita 6, 619334
NEFH	96,3	88,2	100	100	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
OPA1	99,5	96,7	100	99,9	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
ORAI1	99,3	97,1	99,4	96,7	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
PABPN1	64,8	54,7	100	99,2	Oculopharyngeal muscular dystrophy, 164300
PFKM	100	99,7	100	100	Glycogen storage disease VII, 232800
PGAM2	100	100	100	100	Glycogen storage disease X, 261670
PGK1	90,3	73,2	100	100	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94,2	94,1	94,2	94,2	Congenital disorder of glycosylation, type It, 614921

PHKA1	97,8	93,4	100	99,6	Muscle glycogenosis, 300559
PHOX2A	92,8	74,4	100	100	Fibrosis of extraocular muscles, congenital, 2, 602078
PIEZO2	99,8	99,2	100	100	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PIP5K1C	99,2	96,7	99,9	99,2	Lethal congenital contractural syndrome 3, 611369
PLEC	100	99,9	100	100	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950 Epidermolysis bullosa simplex with pyloric atresia, 612138 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with muscular dystrophy, 226670
PNPLA2	99,8	96,1	100	100	Neutral lipid storage disease with myopathy, 610717
POLG	99,9	98,8	100	100	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	99,3	97,2	100	99,8	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	99,8	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 (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
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	99,5	97,3	100	100	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	99,8	97,3	100	100	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	98,9	96,6	100	100	Myasthenic syndrome, congenital, 22, 616224

PRPS1	86,4	86,3	100	99,7	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	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PYGM	100	100	100	100	McArdle disease, 232600
RAPSN	100	99,6	100	100	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	99,9	98,3	100	100	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RRM2B	100	99,8	100	99,9	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RXYLT1	99,2	95,9	100	99,9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	97,1	94	99,4	99	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320
SCN4A	99,9	99,4	100	100	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	84,3	84	87,8	85,1	Myopathy, congenital, with fiber-type disproportion, 255310 Muscular dystrophy, rigid spine, 1, 602771
SGCA	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	97,8	96,5	100	99,9	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	99,6	96,5	100	100	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100	99,4	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SIL1	98,7	96	100	100	Marinesco-Sjogren syndrome, 248800
SLC25A4	100	99,8	100	100	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	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707

SLC52A3	100	100	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SMCHD1	99,3	96,4	100	99,9	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMDT1	100	100	100	100	No OMIM disease ID
SPEG	97,2	91,1	99,7	99,7	Centronuclear myopathy 5, 615959
SRPK3	99	95,7	100	100	No OMIM disease ID
STIM1	99,9	97,5	100	100	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
SYT2	100	98,8	100	100	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TCAP	100	100	100	100	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TK2	99	96	100	100	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TNNI2	100	99,9	100	100	Arthrogryposis, distal, type 2B1, 601680
TNNT1	99,6	97,1	100	100	Nemaline myopathy 5, Amish type, 605355
TNPO3	100	99,8	100	100	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TPM2	100	99,8	100	99,9	Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	87,7	84,3	100	100	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRAPPC11	99,7	98,7	100	99,9	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRIM32	100	99,9	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIP4	99,8	99	100	100	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRPV4	100	99,9	100	100	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	83,8	74,1	100	99,8	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	98,5	98	100	100	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	98,5	96,8	100	100	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	100	99,9	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	99,2	97,3	99,9	99,3	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
VCP	100	99,1	100	100	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	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	99	92,6	100	99,8	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	99,4	97,8	100	100	Pontocerebellar hypoplasia type 1A, 607596
VWA1	85,4	77,7	99,6	96,1	Neuropathy, hereditary motor, with myopathic features, 619216
XK	99,7	97,6	100	100	McLeod syndrome with or without chronic granulomatous disease, 300842
YARS2	99,9	99,4	100	100	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZC4H2	100	98,1	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.
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-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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors