MUSCLE DISORDERS PANEL DG-5.1.0 (228 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ACADVL	100%	100%	100%	100%	99.3%	VLCAD deficiency, 201475
ACTA1	100%	100%	100%	99.8%	97.9%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800
ACTN2	99.8%	98.4%	100%	99.8%	98.6%	Myopathy, distal, 6, adult onset, 618655;Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158;Congenital myopathy 8, 618654;Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158
ACVR1	100%	100%	100%	100%	99.7%	Fibrodysplasia ossificans progressiva, 135100
ADSS1	100%	100%	100%	99.9%	99%	Myopathy, distal, 5, 617030
AGL	100%	100%	100%	100%	99.7%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400
AGRN	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120

ALG2	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
AMPD1	100%	100%	100%	100%	99.7%	Myopathy due to myoadenylate deaminase deficiency, 615511
ANO5	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260
ASCC1	86.6%	86.6%	100%	100%	99.6%	Spinal muscular atrophy with congenital bone fractures 2, 616867;Barrett esophagus/esophageal adenocarcinoma, 614266
ATP2A1	100%	100%	100%	100%	99.3%	Brody myopathy, 601003
ATP2A2	100%	100%	100%	100%	99.6%	Acrokeratosis verruciformis, 101900;Darier disease, 124200;{Rhabdomyolysis, susceptibility to, 2}, 621236
ATP7A	95%	94.8%	99.4%	92.2%	74.7%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
B3GALNT2	92.6%	92.6%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181

B4GAT1	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	100%	100%	100%	100%	99.5%	?Neuronopathy, distal hereditary motor, autosomal dominant 15, 621094;Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954;Charcot-Marie-Tooth disease, axonal, type 2JJ, 621095
BICD2	100%	100%	100%	100%	99.4%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	100%	100%	100%	100%	99.3%	Centronuclear myopathy 2, 255200
CACNA1S	100%	100%	100%	100%	99.4%	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580;Congenital myopathy 18 due to dihydropyridine receptor defect, 620246;Hypokalemic periodic paralysis, type 1, 170400;{Malignant hyperthermia susceptibility 5}, 601887
CAPN3	100%	100%	100%	100%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600;Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129

CASQ1	100%	100%	100%	100%	99.4%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CAV3	100%	100%	100%	100%	99.2%	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%	99.6%	97.4%	Lipodystrophy, congenital generalized, type 4, 613327
CCDC78	100%	100%	100%	100%	99.5%	Centronuclear myopathy 4, 614807
CFL2	100%	100%	100%	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	100%	100%	100%	100%	99.5%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHKB	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462;Multiple pterygium syndrome, lethal type, 253290

CHRNB1	100%	100%	100%	99.8%	98.5%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314;Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	100%	100%	100%	100%	99.5%	?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%	99.9%	98.9%	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%	99.9%	99.6%	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000
CLCN1	100%	100%	100%	100%	99.6%	Myotonia congenita, recessive, 255700;Myotonia congenita, dominant, 160800;Myotonia levior, 160800
CNTN1	100%	100%	100%	100%	99.8%	Congenital myopathy 12, 612540

COL12A1	100%	100%	100%	100%	99.8%	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 19, 616720
COL4A1	100%	100%	100%	100%	99.7%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780
COL6A1	100%	100%	100%	100%	99.4%	Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810
COL6A2	100%	99.8%	100%	99.9%	99.2%	?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725
COL6A3	100%	100%	100%	100%	99.7%	Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411
COLQ	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 5, 603034
COX6A2	100%	100%	100%	99.9%	97.6%	Mitochondrial complex IV deficiency, nuclear type 18, 619062

CPT2	97.3%	96.3%	100%	100%	99.7%	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110
CRPPA	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRYAB	100%	100%	100%	100%	99.7%	Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
DAG1	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS2	100%	100%	100%	100%	99.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485

DES	92.6%	92.6%	100%	99.9%	98.6%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DGUOK	100%	100%	100%	100%	99.9%	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	99.6%	99.6%	99.2%	91.5%	73.3%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DNA2	97.1%	97.1%	100%	100%	99.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807
DNAJB4	100%	100%	100%	100%	99.9%	Congenital myopathy 21 with early respiratory failure, 620326
DNAJB6	92.2%	92.2%	100%	100%	99.9%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511

DNM2	93.3%	91.7%	100%	99.9%	99.4%	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	100%	100%	100%	99.9%	99.3%	Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300
DPAGT1	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
DPM1	99.6%	96.9%	99.6%	98.4%	96.6%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	99.9%	100%	100%	99.5%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100%	100%	100%	100%	98.8%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DYNC1H1	99.3%	99.3%	100%	99.9%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563

DYSF	96.9%	96.9%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601;Miyoshi muscular dystrophy 1, 254130;Myopathy, distal, with anterior tibial onset, 606768
ECEL1	100%	100%	100%	100%	99.3%	Arthrogryposis, distal, type 5D, 615065
EMD	93.1%	91.5%	97.5%	87.2%	69.2%	Cardiomyopathy, dilated, 3C, 301163;Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	100%	100%	100%	100%	98.9%	Glycogen storage disease XIII, 612932
ERBB3	100%	100%	100%	99.9%	98.9%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
EXOSC8	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	100%	100%	100%	100%	99.4%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FDX2	100%	100%	100%	100%	99.3%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900

FHL1	100%	100%	99.4%	91.2%	73.3%	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%	100%	100%	100%	99.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), 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	95.2%	94.9%	100%	100%	99.9%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152
FLAD1	100%	100%	100%	100%	99.5%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLNC	100%	100%	100%	100%	99.6%	Cardiomyopathy, familial dilated, 1PP, 617047;Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524
FXR1	100%	100%	100%	99.9%	99.4%	Congenital myopathy 9B, proximal, with minicore lesions, 618823;?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822
GAA	100%	100%	100%	100%	99.5%	Pompe disease, late-onset, 621314;Pompe disease, infantile-onset, 232300

GATM	100%	100%	100%	100%	99.9%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600
GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GFPT1	97.5%	97.5%	100%	100%	99.8%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GMPPB	100%	100%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GNE	100%	100%	100%	100%	99.8%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820
GRIN1	100%	100%	100%	100%	99.2%	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	93.7%	93.7%	100%	100%	100%	?Glycogen storage disease XV, 613507;Polyglucosan body myopathy 2, 616199
GYS1	100%	100%	100%	100%	99.5%	Glycogen storage disease 0, muscle, 611556
HACD1	71.3%	71.3%	100%	100%	99.6%	Congenital myopathy 11, 619967
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HMGCR	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375;[Statins, response to], 620410;[Low density lipoprotein cholesterol level QTL 3], 620410
HMGCS1	97.2%	97.2%	100%	100%	99.7%	Congenital myopathy 28 with rigid spine, 621433
HNRNPA1	100%	100%	100%	100%	99.7%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424;?Myopathy, distal, 3, 610099;Amyotrophic lateral sclerosis 20, 615426

HNRNPA2B1	100%	100%	100%	100%	99.7%	Oculopharyngeal muscular dystrophy 2, 620460;?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPDL	100%	100%	100%	99.7%	97.5%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HSPG2	100%	100%	100%	100%	99.4%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320
INPP5K	100%	98.5%	100%	100%	99.8%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
ISCU	100%	100%	100%	100%	99.7%	Myopathy with lactic acidosis, hereditary, 255125
ITGA7	100%	99.9%	100%	100%	99.5%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
JAG2	100%	100%	100%	100%	99.1%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
KBTBD13	100%	100%	100%	100%	99.1%	Nemaline myopathy 6, autosomal dominant, 609273

KCNJ2	100%	100%	100%	100%	99.8%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KIF21A	100%	100%	100%	100%	99.6%	Fibrosis of extraocular muscles, congenital, 3B, 135700;Fibrosis of extraocular muscles, congenital, 1, 135700
KLHL40	100%	100%	100%	99.9%	99.5%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	100%	100%	100%	99.7%	Nemaline myopathy 9, 615731
KLHL9	100%	100%	100%	100%	99.7%	
LAMA2	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMP2	70.7%	70.7%	98.9%	91%	71.6%	Danon disease, 300257
LARGE1	96.9%	96.9%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154

LDB3	95.9%	94.9%	100%	100%	99.4%	Left ventricular noncompaction 3, 601493;Cardiomyopathy, dilated, 2L, 621237;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LDHA	100%	100%	100%	100%	99.6%	Glycogen storage disease XI, 612933
LMNA	100%	100%	100%	99.9%	99.3%	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%	100%	100%	99.9%	99.2%	Nemaline myopathy 10, 616165
LPIN1	94%	94%	100%	100%	99.7%	Myoglobinuria, acute recurrent, autosomal recessive, 268200

MAP3K20	100%	100%	100%	100%	99.9%	Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890
MB	100%	100%	100%	100%	99.4%	Myopathy, sarcoplasmic body, 620286
MEGF10	100%	100%	100%	100%	99.6%	Congenital myopathy 10A, severe variant, 614399;Congenital myopathy 10B, mild variant, 620249
MICU1	86.7%	86.7%	100%	100%	99.8%	Myopathy with extrapyramidal signs, 615673
MLIP	100%	100%	100%	100%	99.7%	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138
MSTN	100%	100%	100%	100%	100%	?Muscle hypertrophy, 614160
MSTO1	100%	100%	99.9%	99.1%	97.7%	Myopathy, mitochondrial, and ataxia, 617675
MTM1	100%	100%	99.2%	92.2%	74.7%	Myopathy, centronuclear, X-linked, 310400
MUSK	100%	100%	100%	100%	99.7%	Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYBPC1	100%	100%	100%	100%	99.8%	Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335

MYH2	100%	100%	100%	99.9%	99.8%	Congenital myopathy 6 with ophthalmoplegia, 605637
MYH3	100%	100%	100%	100%	99.5%	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%	99.1%	100%	100%	99.7%	Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358
MYL1	100%	100%	100%	100%	99.7%	Congenital myopathy 14, 618414
MYO18B	100%	100%	100%	99.9%	99.4%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYOD1	100%	100%	100%	99.9%	97.5%	Congenital myopathy 17, 618975
MYOT	100%	100%	100%	100%	99.8%	Myopathy, myofibrillar, 3, 609200

MYPN	98.4%	98.4%	100%	100%	99.6%	Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248
NEB	99.9%	99.8%	99.6%	99.2%	98.3%	Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334
NEFH	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924;{?Amyotrophic lateral sclerosis, susceptibility to}, 105400
OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
ORAI1	100%	100%	100%	99.8%	98.6%	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883
PABPN1	100%	100%	100%	100%	98.8%	Oculopharyngeal muscular dystrophy, 164300

PAX7	94.2%	94.2%	100%	99.9%	99.1%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220
PFKM	100%	100%	100%	100%	99.7%	Glycogen storage disease VII, 232800
PGAM2	100%	100%	100%	99.8%	99.4%	Glycogen storage disease X, 261670
PGK1	92.7%	92.7%	99.3%	90.9%	71.5%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PHKA1	100%	100%	99.5%	91.9%	72.6%	Muscle glycogenosis, 300559
PHOX2A	100%	100%	100%	99.9%	99.2%	Fibrosis of extraocular muscles, congenital, 2, 602078
PIEZO2	100%	100%	100%	100%	99.7%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PIP5K1C	90.6%	90.2%	100%	99.9%	99.4%	Lethal congenital contractural syndrome 3, 611369

PLEC	100%	100%	100%	100%	98.9%	?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	80.4%	80.4%	100%	100%	99.6%	Neutral lipid storage disease with myopathy, 610717
POGLUT1	99.9%	98%	100%	100%	99.8%	Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLG	100%	100%	100%	100%	99.8%	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	95.8%	95.8%	100%	100%	99.5%	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%	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151;Retinitis pigmentosa 76, 617123;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	100%	100%	100%	100%	99.5%	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%	99.8%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249

POMT1	100%	99.7%	100%	100%	99.3%	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 impaired intellectual development), type B, 1, 613155
POMT2	94.9%	94.9%	100%	99.8%	98.4%	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 impaired intellectual development), type B, 2, 613156
POPDC1	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
POPDC3	100%	100%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848
PREPL	100%	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 22, 616224
PRPS1	100%	100%	99%	89.8%	71.8%	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%	99.9%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	100%	100%	100%	99.9%	99.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYGM	100%	100%	100%	100%	99.6%	McArdle disease, 232600
PYROXD1	92.6%	92.6%	100%	100%	99.6%	Myopathy, myofibrillar, 8, 617258
RAPSN	100%	100%	100%	100%	99.6%	Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	100%	100%	100%	99.8%	98.8%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RNASEH1	88.3%	88.3%	100%	100%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM1	100%	100%	100%	100%	99.7%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647

RRM2B	100%	100%	100%	100%	99.9%	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%	100%	100%	99.8%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	100%	100%	100%	99.9%	99%	Congenital myopathy 1B, autosomal recessive, 255320;Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000;King-Denborough syndrome, 619542;{Malignant hyperthermia susceptibility 1}, 145600

SCN4A	100%	100%	100%	100%	99.4%	Paramyotonia congenita, 168300;Hyperkalemic periodic paralysis, 170500;Congenital myopathy 22B, severe fetal, 620369;Hypokalemic periodic paralysis, type 2, 613345;Myotonia congenita, atypical, acetazolamide-responsive, 608390;Myasthenic syndrome, congenital, 16, 614198;Congenital myopathy 22A, classic, 620351
SECISBP2	100%	100%	100%	100%	99.7%	Thyroid hormone metabolism, abnormal, 1, 609698
SELENON	93.9%	91.8%	100%	99.8%	99.3%	Congenital myopathy 3 with rigid spine, 602771
SGCA	97.5%	97.5%	100%	100%	99.2%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94%	94%	100%	100%	99.7%	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	83.2%	83.2%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SH3TC2	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353
SIL1	91.5%	91.5%	100%	100%	99.5%	Marinesco-Sjogren syndrome, 248800

SLC18A3	100%	100%	100%	100%	98.8%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC25A1	100%	100%	100%	99.9%	98.8%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A4	100%	100%	100%	100%	99.4%	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%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A7	100%	100%	100%	99.9%	99.4%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SMCHD1	100%	100%	100%	100%	99.8%	Facioscapulohumeral muscular dystrophy 2, digenic, 158901;Bosma arhinia microphthalmia syndrome, 603457
SMDT1	100%	100%	100%	100%	99.9%	

SNUPN	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793
SPEG	100%	100%	100%	99.9%	98.7%	Centronuclear myopathy 5, 615959
SRPK3	100%	99.9%	97.7%	87.1%	69.3%	Intellectual developmental disorder, X-linked 114, 301134
STAC3	100%	100%	100%	100%	99.2%	Congenital myopathy 13, 255995
STIM1	100%	99.9%	100%	99.9%	99.7%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SYT2	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040;Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
TANGO2	100%	100%	100%	100%	99.6%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TCAP	100%	100%	100%	100%	99.7%	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954

TIA1	92.7%	92.7%	100%	100%	99.8%	Welander distal myopathy, 604454;Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133
TK2	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TNNC2	100%	100%	100%	99.9%	99.1%	Congenital myopathy 15, 620161
TNNI2	100%	100%	100%	100%	99.1%	Arthrogryposis, distal, type 2B1, 601680
TNNT1	99.9%	97.6%	100%	99.9%	98.5%	Nemaline myopathy 5C, autosomal dominant, 620389;Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355;Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386
TNPO3	97.1%	97.1%	100%	100%	99.9%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6B	100%	100%	100%	100%	99.6%	Global developmental delay with speech and behavioral abnormalities, 619243
TOR1AIP1	100%	100%	100%	100%	99.8%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072

TPM2	100%	100%	100%	100%	99.5%	Arthrogryposis, distal, type 2B4, 108120;Arthrogryposis, distal, type 1A, 108120;Congenital myopathy 23, 609285
TPM3	100%	100%	100%	99.9%	99.8%	Congenital myopathy 4A, autosomal dominant, 255310;Congenital myopathy 4B, autosomal recessive, 609284
TRAPPC11	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRDN	92.7%	92.7%	100%	100%	99.8%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TRIM32	100%	100%	100%	100%	99.6%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIP4	95.9%	94.9%	100%	100%	99.7%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066;Spinal muscular atrophy with congenital bone fractures 1, 616866

TRPV4	100%	100%	100%	99.9%	99.1%	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;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;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500
TTC19	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;?Cardiomyopathy, familial hypertrophic, 9, 613765;Myopathy myofibrillar, 9, with early respiratory failure, 603689

TUBA4A	100%	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 23, 621231;Spastic ataxia 11, autosomal dominant, 621226;Frontotemporal dementia and/or amyotrophic lateral sclerosis 9, 616208;Congenital myopathy 26, 621225
TUBB3	100%	100%	100%	100%	99.5%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
UBA1	96.3%	95.6%	99%	90%	71.9%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
UNC45B	96.1%	95.9%	100%	100%	99.7%	?Cataract 43, 616279;Myofibrillar myopathy 11, 619178
VAMP1	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600

VCP	100%	99.6%	100%	100%	99.8%	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	99.8%	97.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	100%	100%	98.5%	86.5%	67.4%	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	99.6%	98%	100%	100%	99.9%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
VWA1	100%	100%	100%	100%	99.1%	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216
XK	100%	99.9%	99.4%	92.2%	74%	McLeod syndrome, 300842
YARS2	100%	100%	100%	100%	99.8%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZC4H2	100%	100%	98.3%	88.3%	72.7%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors