MUSCLE DISORDERS GENE PANEL DG 2.11 (151 genes)

Gene	*Median*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACADVL	118.9	98	95	VLCAD deficiency, 201475
ACTA1	100	99	95	Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ACVR1	165.2	100	100	Fibrodysplasia ossificans progressiva, 135100
AGL	145.9	99	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	115.1	95	89	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	142.1	99	96	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ATP2A1	156.1	100	100	Brody myopathy, 601003
ATP7A	134.1	99	97	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
B3GALNT2	115	92	89	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GNT1	120.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 13, 615287
BAG3	136.9	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	159	100	99	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	100.3	99	95	Myopathy, centronuclear, autosomal recessive, 255200
CACNA1S	135.7	100	99	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CAPN3	109.9	98	96	Muscular dystrophy, limb-girdle, type 2A, 253600
CASQ1	122.1	100	99	Myopathy, vacuolar, with CASQ1 aggregates, 616231

CAV3	305.2	100	100	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Muscular dystrophy, limb-girdle, type IC, 607801 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072
CCDC78	115	100	100	Myopathy, centronuclear, 4, 614807
CFL2	117.3	94	86	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	129.9	89	86	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	20	43	35	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHKB	98.7	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	128.6	100	99	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNB1	132.1	98	96	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314
CHRND	150.9	100	99	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323
CHRNE	127.7	99	95	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CLCN1	137.5	100	99	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive
CNTN1	151.6	99	98	?Myopathy, congenital, Compton-North, 612540
COL12A1	137.6	99	97	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	85.1	99	97	Myasthenic syndrome, congenital, 19, 616720
COL6A1	137.3	99	97	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090

COL6A2	165.5	99	98	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600
COL6A3	174.9	100	99	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COLQ	117	99	98	Myasthenic syndrome, congenital, 5, 603034
CPT2	166.7	98	97	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CRYAB	125.9	99	98	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
DAG1	221.1	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DES	121	99	98	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DMD	113.7	99	97	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DNA2	132	99	97	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAJB6	58.4	90	77	Muscular dystrophy, limb-girdle, type 1E, 603511
DNM2	127.9	97	94	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150
DOK7	99	94	92	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence, 208150
DPAGT1	113	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750

DPM1	131.2	91	86	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.2	100	99	Congenital disorder of glycosylation, type Iu, 615042
DPM3	184.3	100	100	Congenital disorder of glycosylation, type Io, 612937
DYNC1H1	180	100	99	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYSF	133.2	100	99	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	100.9	88	83	Arthrogryposis, distal, type 5D, 615065
EMD	101.1	99	97	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	179.5	100	100	?Glycogen storage disease XIII, 612932
ERBB3	139.5	100	99	Lethal congenital contractural syndrome 2, 607598
EXOSC8	79.9	91	76	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	152.8	100	99	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FHL1	87.8	98	93	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Hemophagocytic lymphohistiocytosis, familial, 1 Myopathy, X-linked, with postural muscle atrophy, 300696 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy, X-linked dominant, 300695
FKBP14	74	100	99	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557
FKRP	94.6	100	99	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	119.9	99	94	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNC	165.3	100	99	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
GAA	128.7	100	99	Glycogen storage disease II, 232300

GBE1	145.4	99	97	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	144.4	99	97	Myasthenia, congenital, 12, with tubular aggregates, 610542
GMPPB	229.1	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNE	153.9	100	99	Nonaka myopathy, 605820 Sialuria, 269921
GYG1	157.9	100	99	Polyglucosan body myopathy 2, 616199 ?Glycogen storage disease XV, 613507
GYS1	110.9	100	98	Glycogen storage disease 0, muscle, 611556
HSPG2	121.4	99	98	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	107.9	99	96	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
ISCU	111.2	100	99	Myopathy with lactic acidosis, hereditary, 255125
ISPD	104.2	95	84	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITGA7	130.4	99	97	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	107.1	99	96	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	229.3	100	100	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KLHL40	158.2	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	203.9	100	99	Nemaline myopathy 9, 615731
KLHL9	260.8	100	100	No OMIM phenotype Myopathy, distal, early-onset (Cirak (2010) Brain 133, 2123)
LAMA2	143.6	99	99	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMP2	106.8	92	91	Danon disease, 300257
LARGE	142.6	100	99	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840

LDB3	127.4	95	93	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	59.8	94	87	Glycogen storage disease XI, 612933
LMNA	89.4	98	91	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, AD, 181350 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210
LMOD3	141.7	99	98	Nemaline myopathy 10, 616165
LPIN1	131.7	99	95	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MB	154.4	100	100	No OMIM phenotype
MEGF10	154.5	100	99	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MICU1	134.1	96	89	Myopathy with extrapyramidal signs, 615673
MSTN	161.2	100	99	Muscle hypertrophy, 614160
MTM1	92.6	98	91	Myotubular myopathy, X-linked, 310400
MUSK	158.5	100	100	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYF6	121.6	100	100	Myopathy, centronuclear, 3, 614408
MYH2	129.4	99	98	Proximal myopathy and ophthalmoplegia, 605637
MYH3	110.4	99	98	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110

MYH7	111.5	99	96	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Liang distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYOT	139.4	99	95	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
NEB	124	82	81	Nemaline myopathy 2, autosomal recessive, 256030
OPA1	122.5	99	94	Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type),616896 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	226.8	92	89	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
PABPN1	66.6	62	60	Oculopharyngeal muscular dystrophy, 164300
PFKM	150.7	100	99	Glycogen storage disease VII, 232800
PGAM2	170.9	100	99	Glycogen storage disease X, 261670
PGK1	54.9	93	81	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	133.7	100	99	Congenital disorder of glycosylation, type It, 614921
PHKA1	107.4	98	95	Muscle glycogenosis, 300559
PIP5K1C	107.7	96	95	Lethal congenital contractural syndrome 3, 611369
PLEC	114.3	99	98	Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, type 2Q, 613723 ?Epidermolysis bullosa simplex with nail dystrophy, 616487
PNPLA2	113.3	99	97	Neutral lipid storage disease with myopathy, 610717
POMGNT1	126.8	99	97	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMGNT2	259.9	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830

POMK	205.3	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094
POMT1	156	99	98	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	111.2	98	97	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PRPS1	150.5	100	100	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PTRF	137.5	99	99	Lipodystrophy, congenital generalized, type 4, 613327
PYGM	127.3	100	99	McArdle disease, 232600
RAPSN	129.5	99	96	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	104.3	99	94	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RRM2B	128.8	99	97	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RYR1	120.9	96	93	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
SCN4A	214.2	99	99	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SEPN1	111.9	85	83	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	144.8	100	99	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	154.1	96	94	Muscular dystrophy, limb-girdle, type 2E, 604286

SGCD	94.8	100	99	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCG	138.9	100	100	Muscular dystrophy, limb-girdle, type 2C, 253700
SLC52A2	177.8	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.7	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SMCHD1	90.8	98	91	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SPEG	91.4	92	85	Centronuclear myopathy 5, 615959
STIM1	127.2	100	99	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070
TANGO2	145.6	100	100	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TCAP	89.3	100	99	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TMEM5	120.5	96	92	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TNNI2	122.5	100	99	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNT1	87.1	96	94	Nemaline myopathy 5, Amish type, 605355
TNPO3	139.7	100	99	Muscular dystrophy, limb-girdle, type 1F, 608423
TPM2	109.4	100	99	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	98.9	89	89	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRAPPC11	126.2	99	96	Muscular dystrophy, limb-girdle, type 2S, 615356
TRIM32	141.3	100	100	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988

TRPV4	172.7	99	98	Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TTC19	93.2	85	79	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	187.7	98	97	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, early-onset, with fatal cardiomyopathy, 611705 Myopathy, proximal, with early respiratory muscle involvement, 603689 Tibial muscular dystrophy, tardive, 600334
UBA1	163.3	99	98	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	144.9	99	99	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VIPAS39	144.7	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	42.4	95	81	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	124.8	97	94	Pontocerebellar hypoplasia type 1A, 607596
XK	97.4	99	99	McLeod syndrome with or without chronic granulomatous disease, 300842
ZC4H2	76.5	99	97	Wieacker-Wolff syndrome, 314580

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 14th, 2017.
This list is accurate for panel version DG 2.11
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors