MUSCLE DISORDERS GENE PANEL DG 2.9

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACADVL	127.9	99%	97%	VLCAD deficiency, 201475
ACTA1	104	99%	97%	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	184.8	100%	99%	Fibrodysplasia ossificans progressiva, 135100
AGL	184.8	99%	99%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGRN	130.4	96%	91%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	168	99%	98%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ATP2A1	176.4	100%	100%	Brody myopathy, 601003
ATP7A	148.2	99%	98%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
B3GALNT2	141.5	93%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GNT1	125.5	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 13, 615287
BAG3	140.5	100%	99%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	162.1	100%	99%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	120.9	99%	97%	Myopathy, centronuclear, autosomal recessive, 255200
CACNA1S	155.5	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CAPN3	126.3	99%	98%	Muscular dystrophy, limb-girdle, type 2A, 253600
CASQ1	128	100%	99%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CAV3	294	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	120.3	100%	99%	Myopathy, centronuclear, 4, 614807
CFL2	134.5	95%	87%	Nemaline myopathy 7, autosomal recessive, 610687
CHAT	146.3	92%	87%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	27.8	65%	42%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHKB	102.6	99%	97%	Muscular dystrophy, congenital, megaconial type, 602541
CHRNA1	134.9	100%	99%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNB1	163.8	99%	97%	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314
CHRND	185.4	99%	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	140.3	99%	97%	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	163.7	100%	99%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive
CNTN1	181.9	99%	98%	?Myopathy, congenital, Compton-North, 612540
COL12A1	167.3	99%	98%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	96.6	99%	96%	Myasthenic syndrome, congenital, 19, 616720
COL6A1	160.6	99%	99%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	179.4	100%	99%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090

				?Myosclerosis, congenital, 255600
COL6A3	201.8	100%	99%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COLQ	123.1	99%	99%	Myasthenic syndrome, congenital, 5, 603034
CPT2	182	98%	96%	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	120.6	99%	96%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
DAG1	236.1	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	145.8	99%	99%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DMD	140.4	99%	98%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DNA2	161.2	99%	99%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAJB6	68.1	91%	78%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNM2	145	99%	95%	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	93.6	94%	92%	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence, 208150
DPAGT1	119.1	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	149.2	92%	87%	Congenital disorder of glycosylation, type Ie, 608799

DPM2	99	99%	96%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	181.4	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DYNC1H1	189.4	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	155.1	100%	99%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768
ECEL1	108.1	90%	82%	Arthrogryposis, distal, type 5D, 615065
EMD	117.4	98%	94%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENO3	204.6	100%	100%	?Glycogen storage disease XIII, 612932
ERBB3	145.7	100%	99%	Lethal congenital contractural syndrome 2, 607598
EXOSC8	102.4	93%	85%	Pontocerebellar hypoplasia, type 1C, 616081
FAM111B	185.5	100%	99%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FHL1	98.4	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	96.5	99%	99%	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557
FKRP	103.2	100%	100%	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	155.6	99%	95%	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	188.5	100%	99%	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
GAA	136.8	100%	99%	Glycogen storage disease II, 232300

GBE1	189.8	99%	98%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GFPT1	171.9	99%	99%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GNE	167.4	100%	99%	Nonaka myopathy, 605820 Sialuria, 269921
GYG1	168.1	100%	99%	Polyglucosan body myopathy 2, 616199 ?Glycogen storage disease XV, 613507
GYS1	120.2	99%	98%	Glycogen storage disease 0, muscle, 611556
HSPG2	133.9	99%	98%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IGHMBP2	123	98%	95%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
ISCU	143.3	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	130.8	97%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITGA7	145.6	99%	96%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	118.6	99%	97%	Nemaline myopathy 6, autosomal dominant, 609273
KCNJ2	210.8	100%	100%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KLHL40	160.3	100%	100%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	228.9	100%	99%	Nemaline myopathy 9, 615731
KLHL9	288.7	100%	100%	No OMIM phenotype Myopathy, distal, early-onset (Cirak (2010) Brain 133, 2123)
LAMA2	176.7	100%	99%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMP2	120.4	93%	91%	Danon disease, 300257
LARGE	142.4	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	146.5	95%	93%	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	69.6	94%	89%	Glycogen storage disease XI, 612933

LMNA	95.6	97%	90%	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	197.4	100%	99%	Nemaline myopathy 10, 616165
LPIN1	151.2	99%	96%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
MEGF10	172.9	100%	100%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MICU1	134.3	97%	93%	Myopathy with extrapyramidal signs, 615673
MSTN	215.3	99%	99%	Muscle hypertrophy, 614160
MTM1	108.7	98%	93%	Myotubular myopathy, X-linked, 310400
MUSK	178.8	100%	100%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYF6	130.1	100%	100%	Myopathy, centronuclear, 3, 614408
MYH2	146.3	99%	99%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	123.5	99%	98%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
MYH7	123.2	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	172.4	99%	98%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200

				Myopathy, spheroid body, 182920
NEB	154.8	83%	82%	Nemaline myopathy 2, autosomal recessive, 256030
ORAI1	243	94%	92%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
PABPN1	88.3	66%	61%	Oculopharyngeal muscular dystrophy, 164300
PFKM	165.3	100%	99%	Glycogen storage disease VII, 232800
PGAM2	177.3	100%	100%	Glycogen storage disease X, 261670
PGK1	57.8	92%	80%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	149.5	100%	99%	Congenital disorder of glycosylation, type It, 614921
PHKA1	120.6	99%	95%	Muscle glycogenosis, 300559
PIP5K1C	131.7	96%	94%	Lethal congenital contractural syndrome 3, 611369
PLEC	122.1	99%	99%	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	139.3	99%	96%	Neutral lipid storage disease with myopathy, 610717
POMGNT1	136.7	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	258.7	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	223.3	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	184.3	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	117.5	99%	96%	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	178.9	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	162.4	99%	99%	Lipodystrophy, congenital generalized, type 4, 613327

PYGM	156.7	100%	100%	McArdle disease, 232600
RAPSN	151.1	99%	95%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RBCK1	112	98%	95%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RRM2B	163	99%	98%	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	136.1	98%	95%	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	229.7	100%	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	133.1	86%	83%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	173.2	100%	99%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	192.5	97%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	109.9	99%	98%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCG	141.7	100%	99%	Muscular dystrophy, limb-girdle, type 2C, 253700
SLC18A2	143.4	100%	99%	No OMIM phenotype Parkinsonian disorder, infantile hypotonic (Jacobsen (2016) J Inherit Metab Dis 39,305) ?Intellectual disability, microcephaly, cortical atrophy and dementia (Karaca (2015) Neuron 88, 499) ?Schizophrenia (Gulsuner (2013) Cell 154, 518)
SLC52A2	206.8	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	108.8	99%	99%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SMCHD1	114.1	98%	94%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SPEG	106.8	96%	89%	Centronuclear myopathy 5, 615959
STIM1	144	99%	97%	Immunodeficiency 10, 612783

				Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070
TANGO2	158.8	100%	100%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TCAP	99.8	99%	97%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TMEM5	138.9	96%	93%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TNNI2	135.4	100%	100%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNT1	103.2	99%	95%	Nemaline myopathy 5, Amish type, 605355
TNPO3	163	100%	99%	Muscular dystrophy, limb-girdle, type 1F, 608423
TPM2	120.8	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	96.1	89%	87%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRAPPC11	156	99%	97%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRIM32	147.3	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRPV4	188.6	99%	99%	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	105.8	90%	82%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTN	236.7	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	176.9	99%	98%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VCP	155.7	100%	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	156.9	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VMA21	56.6	96%	87%	Myopathy, X-linked, with excessive autophagy, 310440
VRK1	157.2	99%	97%	Pontocerebellar hypoplasia type 1A, 607596

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 2.9

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors