MUSCLE DISORDERS GENE PANEL

Gene Symbol	Depth (Reads)	Coverage (avg %)	OMIM disease	Description
ACADVL	98	89	201475	VLCAD deficiency
ACTA1	71	55	161800	Myopathy actin congenital with cores
ACVR1	115	84	135100	Fibrodysplasia ossificans progressiva
AGL	134	97	232400	Glycogen storage disease IIIa
AGRN	76	91	254300	Myasthenia limb-girdle familial
ANO5	105	93	166260	Gnathodiaphyseal dysplasia
ATP2A1	115	89	601003	Brody myopathy
BAG3	161	97	613881	Cardiomyopathy dilated 1HH
BICD2	91	96	615290	Spinal muscular atrophy lower extremity-predominant 2 AD
BIN1	52	89	255200	Myopathy centronuclear autosomal recessive
CACNA1S	93	92	170400	Hypokalemic periodic paralysis type 1
CAPN3	128	93	253600	Muscular dystrophy limb-girdle type 2A
CAV3	132	100	192600	Cardiomyopathy familial hypertrophic
CCDC78	84	95	614807	Myopathy centronuclear 4
CFL2	113	100	610687	Nemaline myopathy 7 autosomal recessive
CHAT	71	81	254210	Myasthenic syndrome congenital associated with episodic apnea
CHKB	90	90	602541	Muscular dystrophy congenital megaconial type
CHRNA1	111	82	253290	Multiple pterygium syndrome lethal type
CHRNB1	113	84	608931	Myasthenic syndrome congenital associated with acetylcholine receptor deficiency
CHRND	106	84	253290	Multiple pterygium syndrome lethal type
CHRNE	139	100	608931	Myasthenic syndrome congenital associated with acetylcholine receptor deficiency
CLCN1	96	90	160800	Myotonia congenita dominant
CNTN1	104	94	612540	Myopathy congenital Compton-North
COL6A1	78	93	158810	Bethlem myopathy
COL6A2	80	95	158810	Bethlem myopathy
COL6A3	130	94	158810	Bethlem myopathy
COLQ	85	87	603034	Endplate acetylcholinesterase deficiency
CPT2	116	96	600649	CPT deficiency hepatic type II

CRYAB	125	98	615184	Cardiomyopathy dilated 1II
DAG1	127	100	613818	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 9
DES	94	94	604765	Cardiomyopathy dilated 1I
DMD	91	91	300376	Becker muscular dystrophy
DNAJB6	70	48	603511	Muscular dystrophy limb-girdle type 1E
DNM2	85	89	606482	Charcot-Marie-Tooth disease axonal type 2M
DOK7	55	88	208150	Fetal akinesia deformation sequence
DPAGT1	116	89	608093	Congenital disorder of glycosylation type Ij
DPM2	72	90	615042	Congenital disorder of glycosylation type Iu
DPM3	88	100	612937	Congenital disorder of glycosylation type Io
DYNC1H1	133	88	614228	Charcot-Marie-Tooth disease axonal type 20
DYSF	104	88	254130	Miyoshi muscular dystrophy 1
EMD	127	100	310300	Emery-Dreifuss muscular dystrophy 1 X-linked
ENO3	113	90	612932	Glycogen storage disease XIII
ERBB3	137	87	607598	Lethal congenital contractural syndrome 2
FHL1	66	70	300696	Emery-Dreifuss muscular dystrophy 6 X-linked
FKRP	67	98	613153	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 5
FKTN	113	91	611615	Cardiomyopathy dilated 1X
FLNC	89	92	614065	Myopathy distal 4
GAA	98	91	232300	Glycogen storage disease II
GBE1	109	96	232500	Glycogen storage disease IV
GFPT1	109	96	610542	Myasthenia congenital with tubular aggregates 1
GNE	112	84	600737	Inclusion body myopathy autosomal recessive
GYG1	79	42	613507	Glycogen storage disease XV
GYS1	77	85	611556	Glycogen storage disease 0 muscle
HSPG2	77	87	224410	Dyssegmental dysplasia Silverman-Handmaker type
IGHMBP2	80	85	604320	Neuronopathy distal hereditary motor type VI
ISCU	87	75	255125	Myopathy with lactic acidosis hereditary
ISPD	97	87	614643	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 7
ITGA7	92	89	613204	Muscular dystrophy congenital due to ITGA7 deficiency
KBTBD13	44	99	609273	Nemaline myopathy 6 autosomal dominant
KCNJ2	136	98	170390	Andersen syndrome
KLHL9	154	48	200	-

LAMA2	112	86	607855	Muscular dystrophy congenital merosin-deficient
LAMP2	90	87	300257	Danon disease
LARGE	116	82	613154	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6
LDB3	92	87	601493	Cardiomyopathy dilated 1C
LDHA	66	22	612933	Glycogen storage disease XI
LMNA	80	86	115200	Cardiomyopathy dilated 1A
LPIN1	112	87	268200	Myoglobinuria acute recurrent autosomal recessive
MATR3	148	51	606070	Myopathy distal 2
MEGF10	114	81	614399	Myopathy areflexia respiratory distress and dysphagia early-onset
MSTN	162	95	614160	Muscle hypertrophy
MTM1	89	89	310400	Myotubular myopathy X-linked
MUSK	132	87	608931	Myasthenic syndrome congenital associated with acetylcholine receptor deficiency
MYBPC3	86	85	115197	Cardiomyopathy familial hypertrophic 4
MYF6	135	100	614408	Myopathy centronuclear 3
MYH2	135	52	605637	Inclusion body myopathy-3
MYH3	144	62	193700	Arthrogryposis distal type 2A
MYH7	118	61	613426	Cardiomyopathy dilated 1S
MYOT	137	96	159000	Muscular dystrophy limb-girdle type 1A
NEB	122	85	256030	Nemaline myopathy 2 autosomal recessive
PABPN1	50	45	164300	Oculopharyngeal muscular dystrophy
PFKM	130	80	232800	Glycogen storage disease VII
PGAM2	77	100	261670	Glycogen storage disease X
PGK1	82	43	300653	Phosphoglycerate kinase 1 deficiency
PGM1	109	76	614921	Congenital disorder of glycosylation type It
PHKA1	80	90	300559	Muscle glycogenosis
PIP5K1C	59	91	611369	Lethal congenital contractural syndrome 3
PLEC	101	50	613723	Muscular dystrophy, limb-girdle, type 2Q
PNPLA2	69	91	610717	Neutral lipid storage disease with myopathy
POMGNT1	102	86	253280	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 3
POMGNT2	114	100	614830	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8
POMT1	118	87	236670	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 1
POMT2	84	90	613150	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 2
PTRF	135	97	613327	Lipodystrophy congenital generalized type 4

PYGM	100	86	232600	McArdle disease
RAPSN	95	83	208150	Fetal akinesia deformation sequence
RYR1	86	89	117000	Central core disease
SCN4A	127	93	170500	Hyperkalemic periodic paralysis type 2
SEPN1	80	90	602771	Muscular dystrophy rigid spine 1
SGCA	100	78	608099	Muscular dystrophy limb-girdle type 2D
SGCB	136	97	604286	Muscular dystrophy limb-girdle type 2E
SGCD	108	87	606685	Cardiomyopathy dilated 1L
SGCG	84	99	253700	Muscular dystrophy limb-girdle type 2C
SYNE1	120	87	612998	Emery-Dreifuss muscular dystrophy 4 autosomal dominant
SYNE2	114	90	612999	Emery-Dreifuss muscular dystrophy 5 autosomal dominant
TCAP	57	81	607487	Cardiomyopathy dilated 1N
TNNI2	55	100	601680	Arthrogryposis multiplex congenita distal type 2B
TNNT1	90	94	605355	Nemaline myopathy 5 Amish type
TPM2	102	86	108120	Arthrogryposis multiplex congenita distal type 1
TPM3	97	60	609284	CAP myopathy 1
TRIM32	110	100	209900	Bardet-Biedl syndrome 11
TRPV4	91	92	113500	Brachyolmia type 3
TTN	159	97	604145	Cardiomyopathy dilated 1G
UBA1	107	94	301830	Spinal muscular atrophy X-linked 2 infantile
VCP	131	86	613954	Amyotrophic lateral sclerosis 14 with or without frontotemporal dementia
VIPAS39	113	83	613404	Arthrogryposis renal dysfunction and cholestasis 2

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors