WES MUSCLE DISORDERS DG 3.00

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACADVL	138.0	99.4	97.3	201475
ACTA1	86.4	99.6	92.3	255310
ACVR1	181.9	100.0	100.0	135100
AGL	180.5	100.0	99.4	232400
AGRN	139.7	96.9	92.6	254300
ANO5	156.6	99.5	97.3	611307
ATP2A1	158.2	100.0	100.0	601003
ATP7A	134.6	99.0	96.9	309400
B3GALNT2	117.0	93.8	89.4	615181
B4GAT1	118.1	100.0	100.0	615287
BAG3	184.0	100.0	100.0	612954
BICD2	137.0	100.0	99.7	615290
BIN1	119.1	99.6	95.7	255200
CACNA1S	130.8	100.0	99.9	170400
CAPN3	118.2	97.8	97.2	253600
CASQ1	118.3	100.0	99.5	616231
CAV3	229.2	100.0	100.0	614321;606072;607801
CAVIN1	158.7	100.0	100.0	613327
CCDC78	124.5	100.0	100.0	614807
CFL2	167.1	100.0	99.6	610687
CHAT	127.1	93.5	85.7	254210
CHCHD10	25.2	59.1	43.9	615048
CHKB	114.9	100.0	99.7	602541
CHRNA1	120.4	100.0	99.2	601462
CHRNB1	163.0	100.0	99.4	601462

CHRND	158.8	99.7	97.9	601462
CHRNE	156.9	100.0	100.0	601462
CHRNG	149.9	100.0	100.0	265000
CLCN1	142.2	100.0	99.2	160800;255700
CNTN1	143.1	99.9	98.9	612540
COL12A1	153.4	100.0	99.4	616471
COL13A1	94.7	93.9	93.8	616720
COL6A1	149.2	100.0	99.4	158810;254090
COL6A2	161.0	100.0	99.8	158810;254090;255600
COL6A3	171.8	100.0	99.8	158810;254090
COLQ	113.9	100.0	99.2	603034
CPT2	159.1	98.2	97.8	255110
CRPPA	122.2	98.5	94.8	614643
CRYAB	108.3	100.0	99.2	608810
DAG1	203.4	100.0	100.0	613818
DARS2	143.6	94.9	94.3	611105
DES	119.2	100.0	99.7	601419
DGUOK	124.5	100.0	99.4	617070
DMD	133.1	99.6	98.6	310200;300376
DNA2	155.0	99.8	98.3	615156
DNAJB6	68.6	96.5	88.5	611332
DNM2	138.9	98.1	94.5	160150
DOK7	103.6	95.1	91.6	254300
DPAGT1	90.7	100.0	100.0	614750
DPM1	155.7	98.2	91.3	608799
DPM2	98.8	100.0	98.7	615042
DPM3	225.1	100.0	100.0	612937
DYNC1H1	157.5	99.9	99.4	158600
DYSF	149.9	100.0	99.9	253601;254130
ECEL1	99.8	95.4	90.0	615065

EMD	150.1	99.9	98.4	310300
ENO3	186.4	100.0	99.9	131370
ERBB3	132.9	100.0	99.8	607598
EXOSC8	91.0	97.9	91.2	616081
FAM111B	193.6	100.0	99.9	615704
FHL1	67.0	99.7	95.8	300718;300719;300695;300696
FKBP14	102.2	100.0	99.9	614557
FKRP	124.4	100.0	100.0	236670;253280;606612;607155
FKTN	128.6	99.7	97.0	236670;611588;253800
FLNC	158.4	100.0	99.6	614065;609524
GAA	156.2	100.0	99.9	232300
GATM	156.2	100.0	100.0	612718
GBE1	200.1	100.0	99.6	232500
GFPT1	190.8	100.0	99.4	610542
GMPPB	245.6	100.0	100.0	615350
GNE	132.7	100.0	99.7	600737;605820
GRIN1	164.3	100.0	100.0	614254
GYG1	151.7	99.9	99.2	613507
GYS1	131.5	100.0	98.6	611556
HSPG2	122.2	99.2	97.7	255800;224410
IGHMBP2	118.2	98.8	95.1	604320
INPP5K	109.2	100.0	100.0	617404
ISCU	135.3	100.0	100.0	255125
ITGA7	142.3	99.6	98.0	613204
KBTBD13	102.9	99.8	95.8	609273
KCNJ2	183.9	100.0	100.0	170390
KIF21A	147.7	99.9	99.3	135700
KLHL40	134.3	100.0	100.0	615348
KLHL41	218.5	100.0	99.9	615731
KLHL9	222.5	100.0	100.0	-

LAMA2	155.8	100.0	99.6	607855
LAMP2	108.9	99.2	95.6	300257
LARGE1	129.6	100.0	99.6	608840
LDB3	149.7	95.4	94.7	609452
LDHA	79.1	95.0	91.7	612933
LMNA	97.4	97.4	91.9	159001;151660;613205;605588;181350
LMOD3	159.8	100.0	99.7	616165
LPIN1	147.0	99.6	97.3	605518
MAP3K20	132.4	100.0	99.5	617760
MEGF10	147.8	100.0	100.0	614399
MICU1	126.1	98.9	95.2	615673
MLIP	176.2	99.9	99.0	-
MSTN	203.2	100.0	100.0	614160
MTM1	95.6	99.0	93.3	310400
MUSK	155.4	100.0	99.9	608931
MYH2	130.0	99.9	99.4	605637
MYH3	113.3	99.9	99.0	601680
MYH7	103.2	99.6	97.3	608358;160500
MYOT	174.8	100.0	99.6	182920;159000;609200
MYPN	157.0	100.0	99.7	617336
NEB	125.5	83.0	82.6	256030
NEFH	108.0	93.4	84.5	616924
OPA1	155.2	99.6	97.6	125250
ORAI1	183.1	99.1	96.4	615883
PABPN1	91.8	66.3	56.9	164300
PFKM	135.4	100.0	99.5	232800
PGAM2	168.8	100.0	100.0	261670
PGK1	50.7	92.8	79.3	300653
PGM1	147.7	94.2	94.2	612934
PHKA1	113.5	99.2	95.3	300559

PIEZO2	121.6	100.0	99.5	108145;114300
PIP5K1C	138.1	98.0	95.8	611369
PLEC	115.2	100.0	99.8	613723
PNPLA2	128.9	99.7	96.1	610717
POMGNT1	131.5	100.0	99.9	613157;253280;606822
POMGNT2	186.0	100.0	100.0	614830
POMK	180.7	100.0	100.0	615249
POMT1	150.5	99.3	97.5	609308;236670
POMT2	110.7	99.4	96.4	253280;613158
PREPL	132.5	99.8	98.2	616224
PRPS1	118.4	86.4	86.4	301835
PTRH2	259.0	100.0	100.0	616263
PYGM	138.7	100.0	99.9	232600
RAPSN	162.4	100.0	99.7	608931
RBCK1	105.6	99.9	98.2	615895
RRM2B	163.8	100.0	99.7	612075
RXYLT1	168.9	99.5	96.8	615041
RYR1	126.2	96.9	93.9	117000;255320
SCN4A	187.3	100.0	99.6	170500;168300;608390;613345
SELENON	141.2	84.5	84.0	255310;602771
SGCA	159.4	100.0	99.9	608099
SGCB	184.2	97.7	96.5	604286
SGCD	88.5	100.0	98.9	601287
SGCG	137.6	100.0	99.2	253700
SLC25A4	144.8	100.0	100.0	609283
SLC52A2	170.8	100.0	100.0	614707
SLC52A3	125.5	100.0	100.0	211530
SMCHD1	116.5	99.5	96.3	158901
SMDT1	202.5	100.0	100.0	-
SPEG	122.8	96.1	88.7	615959

SRPK3	119.6	98.7	96.1	-
STIM1	126.2	99.8	98.0	160565
SYT2	98.5	99.9	99.0	-
TANGO2	133.1	100.0	99.3	616878
TCAP	88.8	100.0	100.0	601954
TK2	119.9	99.2	96.3	609560
TNNI2	130.3	100.0	99.7	601680
TNNT1	107.6	99.9	97.6	605355
TNPO3	134.4	100.0	99.9	608423
TPM2	119.2	100.0	100.0	601680;108120;609285
TPM3	85.5	89.2	87.2	609284
TRAPPC11	151.1	100.0	99.2	615356
TRIM32	131.1	100.0	100.0	254110
TRIP4	124.2	100.0	99.1	616866
TRPV4	158.6	100.0	99.9	600175
TTC19	89.7	81.5	73.8	615157
TTN	198.1	98.6	98.1	603689;600334;608807
TUBB3	113.8	98.3	96.9	600638
TWNK	202.8	100.0	100.0	609286
UBA1	142.9	99.4	98.2	301830
VCP	122.7	100.0	99.2	167320
VIPAS39	133.8	100.0	100.0	613404
VMA21	89.1	99.0	94.6	310440
VRK1	156.9	99.7	98.5	607596
XK	83.0	99.8	98.1	300842
YARS2	194.6	100.0	99.8	613561
ZC4H2	87.8	100.0	99.0	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 10x 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 : October 1st, 2016.

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