

WES MUSCLE DISORDERS DG 2.15

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ACADVL	118.8	98.7	95.1	201475
ACTA1	99.7	99.2	95.3	255310
ACVR1	165.1	100.0	100.0	135100
AGL	146.7	99.7	98.0	232400
AGRN	114.8	95.2	89.3	254300
ANO5	142.2	99.5	95.9	611307
ATP2A1	155.9	100.0	100.0	601003
ATP7A	133.2	99.7	97.8	309400
B3GALNT2	115.0	92.4	89.7	615181
B4GAT1	120.4	100.0	100.0	615287
BAG3	136.5	100.0	100.0	612954
BICD2	158.6	100.0	99.9	615290
BIN1	100.2	99.1	95.3	255200
CACNA1S	135.5	100.0	99.7	170400
CAPN3	111.4	99.0	96.7	253600
CASQ1	122.0	100.0	99.6	616231
CAV3	304.7	100.0	100.0	614321;606072;607801
CAVIN1	137.0	99.9	99.3	613327
CCDC78	114.9	100.0	100.0	614807
CFL2	117.3	94.3	86.7	610687
CHAT	130.3	89.3	86.8	254210
CHCHD10	20.0	43.0	35.2	615048
CHKB	98.5	100.0	99.0	602541
CHRNA1	121.8	94.7	94.6	601462
CHRNB1	131.8	98.8	96.7	601462

CHRND	150.5	100.0	99.0	601462
CHRNE	127.7	99.3	95.8	601462
CLCN1	137.4	100.0	99.5	160800;255700
CNTN1	151.7	99.8	98.3	612540
COL12A1	137.5	99.5	97.5	616471
COL13A1	85.4	99.8	97.1	616720
COL6A1	137.2	99.5	97.8	158810;254090
COL6A2	165.3	99.3	98.4	158810;255600;254090
COL6A3	174.7	100.0	99.9	158810;254090
COLQ	113.2	99.8	98.1	603034
CPT2	162.8	97.2	95.4	255110
CRYAB	125.7	99.9	98.7	608810
DAG1	220.8	100.0	100.0	613818
DES	120.8	99.9	98.1	601419
DGUOK	119.2	100.0	100.0	617070
DMD	112.4	99.4	97.4	310200;300376
DNA2	123.6	99.8	96.9	615156
DNAJB6	59.8	91.7	79.3	611332
DNM2	127.4	97.5	94.4	160150
DOK7	105.7	93.3	92.5	254300
DPAGT1	110.7	100.0	100.0	614750
DPM1	131.1	91.7	86.7	608799
DPM2	102.1	100.0	99.4	615042
DPM3	183.9	100.0	100.0	612937
DYNC1H1	179.8	100.0	99.6	158600
DYSF	133.1	100.0	99.8	253601;254130
ECEL1	100.7	88.8	83.1	615065
EMD	100.3	99.8	97.2	310300
ENO3	179.3	100.0	100.0	131370
ERBB3	139.2	100.0	99.9	607598

EXOSC8	80.1	91.8	76.9	616081
FAM111B	152.9	100.0	99.8	615704
FHL1	87.2	98.8	93.0	300696;300718;300695;300719
FKBP14	74.3	100.0	99.4	614557
FKRP	94.5	100.0	99.7	236670;607155;606612;253280
FKTN	120.0	99.2	94.2	236670;611588;253800
FLNC	165.0	100.0	99.7	614065;609524
GAA	128.5	100.0	99.9	232300
GBE1	145.5	99.6	97.2	232500
GFPT1	144.4	99.9	97.6	610542
GMPPB	228.5	100.0	100.0	615350
GNE	153.7	100.0	99.8	600737;605820
GRIN1	150.7	100.0	99.5	614254
GYG1	157.8	100.0	99.6	613507
GYS1	110.8	100.0	98.5	611556
HSPG2	121.3	99.4	98.2	255800;224410
IGHMBP2	107.8	99.3	96.0	604320
INPP5K	108.3	100.0	99.6	617404
ISCU	111.2	100.0	99.7	255125
ISPD	104.4	95.2	84.8	614643
ITGA7	129.6	99.6	97.6	613204
KBTBD13	107.1	99.8	96.8	609273
KCNJ2	229.3	100.0	100.0	170390
KLHL40	157.9	100.0	100.0	615348
KLHL41	203.5	100.0	99.6	615731
KLHL9	260.8	100.0	100.0	1
LAMA2	143.5	99.9	99.5	607855
LAMP2	106.1	92.7	91.2	300257
LARGE1	143.0	100.0	99.6	608840
LDB3	127.3	95.5	93.7	609452

LDHA	59.8	94.1	87.1	612933
LMNA	89.2	97.9	91.3	605588;151660;181350;159001;613205
LMOD3	141.5	99.9	98.5	616165
LPIN1	134.6	99.8	97.8	605518
MB	153.8	100.0	100.0	-
MEGF10	154.3	100.0	99.8	614399
MICU1	134.2	96.0	88.8	615673
MSTN	161.5	100.0	99.5	614160
MTM1	93.7	99.2	93.0	310400
MUSK	159.4	100.0	99.9	608931
MYF6	121.6	100.0	100.0	614408
MYH2	129.4	99.9	98.8	605637
MYH3	110.4	99.9	98.6	601680
MYH7	111.4	99.4	96.8	160500;608358
MYOT	139.4	99.3	95.5	182920;609200;159000
NEB	124.0	82.9	81.9	256030
OPA1	122.5	99.1	94.1	125250
ORAI1	237.3	93.8	89.8	615883
PABPN1	66.6	62.4	60.5	164300
PFKM	150.4	100.0	99.8	232800
PGAM2	170.9	100.0	99.9	261670
PGK1	54.5	93.3	81.6	300653
PGM1	133.6	100.0	99.9	612934
PHKA1	106.7	98.9	95.3	300559
PIP5K1C	107.6	96.3	95.1	611369
PLEC	114.1	99.7	98.7	613723
PNPLA2	113.2	99.7	97.4	610717
POMGNT1	127.6	99.7	97.1	613157;606822;253280
POMGNT2	259.6	100.0	100.0	614830
POMK	205.1	100.0	100.0	615249

POMT1	155.7	99.7	98.1	236670;609308
POMT2	111.1	98.9	97.5	613158;253280
PRPS1	149.5	100.0	100.0	301835
PYGM	127.1	100.0	99.9	232600
RAPSN	140.5	99.6	96.3	608931
RBCK1	104.1	99.2	94.9	615895
RRM2B	128.6	99.7	97.5	612075
RYR1	120.7	96.8	93.7	117000;255320
SCN4A	214.0	99.9	99.5	168300;170500;608390;613345
SELENON	111.7	85.2	83.3	255310;602771
SGCA	147.3	100.0	99.7	608099
SGCB	154.2	96.6	94.2	604286
SGCD	94.8	100.0	99.4	601287
SGCG	138.7	100.0	100.0	253700
SLC25A4	134.1	100.0	100.0	609283
SLC52A2	177.6	100.0	100.0	614707
SLC52A3	119.6	100.0	100.0	211530
SMCHD1	91.3	98.1	92.3	158901
SPEG	100.9	93.3	86.5	615959
STIM1	145.3	100.0	99.2	160565
TANGO2	145.3	100.0	100.0	616878
TCAP	89.0	100.0	99.2	601954
TK2	105.7	93.4	89.4	609560
TMEM5	120.5	96.8	92.9	615041
TNNI2	121.2	100.0	99.6	601680
TNNT1	86.9	96.3	94.0	605355
TNPO3	139.6	100.0	99.7	608423
TPM2	109.1	100.0	99.6	609285;601680;108120
TPM3	98.9	89.4	89.1	609284
TRAPPC11	126.2	99.4	96.4	615356

TRIM32	141.2	100.0	100.0	254110
TRIP4	113.5	100.0	98.8	616866
TRPV4	172.4	99.5	98.7	600175
TTC19	92.1	80.6	72.5	615157
TTN	187.8	98.2	97.2	608807;603689;600334
TWNK	178.8	100.0	100.0	609286
UBA1	162.0	99.8	98.9	301830
VCP	144.8	99.9	99.5	167320
VIPAS39	144.6	100.0	100.0	613404
VMA21	42.3	95.2	81.3	310440
VRK1	124.8	97.5	94.2	607596
XK	96.8	99.9	99.1	300842
YARS2	173.2	99.8	98.9	613561
ZC4H2	78.6	99.8	98.1	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 : 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