

WES MUSCLE DISORDERS DG 3.5

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ACADVL	143.7	100.0	100.0	201475
ACTA1	190.5	100.0	100.0	255310
ACVR1	128.3	100.0	99.9	135100
AGL	158.2	100.0	100.0	232400
AGRN	146.7	100.0	100.0	254300
ANO5	150.3	100.0	100.0	611307
ASCC1	117.4	86.7	86.6	616867
ATP2A1	120.9	100.0	100.0	601003
ATP7A	107.5	100.0	100.0	309400
B3GALNT2	125.3	92.4	92.4	615181
B4GAT1	116.1	100.0	100.0	615287
BAG3	113.6	100.0	100.0	612954
BICD2	119.7	100.0	100.0	615290
BIN1	119.6	100.0	100.0	255200
CACNA1S	121.7	100.0	100.0	170400
CAPN3	134.3	100.0	100.0	253600
CASQ1	114.2	100.0	100.0	616231
CAV3	137.2	100.0	100.0	614321;606072;607801
CAVIN1	119.8	100.0	100.0	613327
CCDC78	149.8	100.0	100.0	614807
CFL2	151.8	100.0	100.0	610687
CHAT	116.7	100.0	100.0	254210
CHCHD10	134.4	100.0	100.0	615048
CHKB	137.9	100.0	100.0	602541
CHRNA1	131.3	100.0	100.0	601462

CHRN1	122.7	100.0	100.0	601462
CHRN2	126.6	100.0	100.0	601462
CHRN3	145.8	100.0	100.0	601462
CHRN4	123.2	100.0	100.0	265000
CLCN1	126.0	100.0	100.0	160800;255700
CNTN1	147.7	100.0	100.0	612540
COL12A1	146.0	100.0	100.0	616471
COL13A1	126.2	100.0	100.0	616720
COL6A1	129.9	100.0	100.0	158810;254090
COL6A2	134.0	100.0	100.0	158810;254090;255600
COL6A3	129.6	100.0	100.0	158810;254090
COLQ	126.6	100.0	100.0	603034
CPT2	139.5	100.0	100.0	255110
CRPPA	154.2	100.0	100.0	614643
CRYAB	133.2	100.0	100.0	608810
DAG1	119.7	100.0	100.0	613818
DARS2	133.3	100.0	100.0	611105
DES	135.4	100.0	100.0	601419
DGUOK	135.8	100.0	100.0	617070
DMD	109.2	99.5	99.1	310200;300376
DNA2	147.9	100.0	100.0	615156
DNAJB6	136.8	100.0	100.0	611332
DNM2	114.9	100.0	100.0	160150
DOK7	126.1	100.0	100.0	254300
DPAGT1	139.6	100.0	100.0	614750
DPM1	143.5	99.2	96.6	608799
DPM2	101.3	100.0	100.0	615042
DPM3	119.4	100.0	100.0	612937
DYNC1H1	125.6	100.0	100.0	158600
DYSF	127.1	100.0	100.0	253601;254130

ECEL1	131.0	100.0	100.0	615065
EMD	88.6	100.0	99.5	310300
ENO3	132.9	100.0	100.0	131370
ERBB3	123.4	100.0	100.0	607598
EXOSC8	147.9	100.0	100.0	616081
FAM111B	161.6	100.0	100.0	615704
FHL1	97.9	100.0	99.9	300718;300719;300695;300696
FKBP14	129.9	100.0	100.0	614557
FKRP	135.4	100.0	100.0	236670;253280;606612;607155
FKTN	163.6	100.0	100.0	236670;611588;253800
FLNC	132.3	100.0	100.0	614065;609524
GAA	128.9	100.0	100.0	232300
GATM	140.0	100.0	100.0	612718
GBE1	156.7	100.0	99.9	232500
GFPT1	140.7	100.0	100.0	610542
GMPPB	122.5	100.0	100.0	615350
GNE	126.6	100.0	100.0	600737;605820
GRIN1	126.3	100.0	100.0	614254
GYG1	132.9	100.0	100.0	613507
GYS1	115.9	100.0	100.0	611556
HADHA	128.3	100.0	100.0	609015
HADHB	137.1	100.0	100.0	609015
HSPG2	131.6	100.0	100.0	255800;224410
IGHMBP2	128.5	100.0	100.0	604320
INPP5K	119.4	100.0	100.0	617404
ISCU	142.2	100.0	100.0	255125
ITGA7	124.6	100.0	100.0	613204
KBTBD13	132.9	100.0	100.0	609273
KCNJ2	153.4	100.0	100.0	170390
KIF21A	149.3	100.0	100.0	135700

KLHL40	126.1	100.0	100.0	615348
KLHL41	149.1	100.0	100.0	615731
KLHL9	155.0	100.0	100.0	-
LAMA2	144.0	99.8	99.5	607855
LAMP2	116.5	100.0	100.0	300257
LARGE1	122.9	100.0	100.0	608840
LDB3	128.3	100.0	100.0	609452
LDHA	139.4	100.0	100.0	612933
LMNA	136.0	100.0	100.0	159001;151660;613205;605588;181350
LMOD3	139.1	100.0	100.0	616165
LPIN1	134.7	100.0	100.0	605518
MAP3K20	138.6	100.0	100.0	617760
MEGF10	129.9	100.0	100.0	614399
MICU1	141.3	100.0	99.9	615673
MLIP	141.6	100.0	100.0	-
MSTN	144.0	100.0	100.0	614160
MSTO1	131.5	100.0	100.0	617675
MTM1	108.2	99.7	99.2	310400
MUSK	131.5	100.0	100.0	608931
MYH2	177.8	100.0	100.0	605637
MYH3	150.8	100.0	100.0	601680
MYH7	152.3	100.0	100.0	608358;160500
MYOT	147.5	100.0	100.0	182920;159000;609200
MYPN	132.1	100.0	100.0	617336
NEB	145.8	99.7	99.2	256030
NEFH	158.6	100.0	100.0	616924
OPA1	148.0	100.0	100.0	125250
ORAI1	123.0	100.0	100.0	615883
PABPN1	144.0	100.0	100.0	164300
PFKM	132.0	100.0	100.0	232800

PGAM2	158.7	100.0	100.0	261670
PGK1	93.9	100.0	99.7	300653
PGM1	127.5	94.0	94.0	612934
PHKA1	107.6	100.0	100.0	300559
PHOX2A	110.5	100.0	100.0	602078
PIEZO2	133.8	100.0	100.0	108145;114300
PIP5K1C	125.9	100.0	100.0	611369
PLEC	137.6	100.0	100.0	613723
PNPLA2	162.3	100.0	100.0	610717
POLG	138.3	100.0	100.0	157640
POLG2	141.0	100.0	100.0	610131
POMGNT1	124.8	100.0	100.0	613157;253280;606822
POMGNT2	117.6	100.0	100.0	614830
POMK	122.5	100.0	100.0	615249
POMT1	132.0	100.0	100.0	609308;236670
POMT2	137.7	100.0	100.0	253280;613158
PREPL	148.2	100.0	100.0	616224
PRPS1	111.3	100.0	100.0	301835
PTRH2	156.6	100.0	100.0	616263
PYGM	125.2	100.0	100.0	232600
PYROXD1	140.1	100.0	100.0	617258
RAPSN	110.4	100.0	100.0	608931
RBCK1	141.8	100.0	100.0	615895
RRM2B	144.5	100.0	100.0	612075
RXYLT1	150.4	100.0	100.0	615041
RYR1	118.9	100.0	99.9	117000;255320
SCN4A	128.7	100.0	100.0	170500;168300;608390;613345
SELENON	108.2	93.1	91.1	255310;602771
SGCA	134.4	100.0	100.0	608099
SGCB	149.2	100.0	100.0	604286

SGCD	142.1	100.0	99.8	601287
SGCG	151.0	100.0	100.0	253700
SIL1	134.7	100.0	100.0	248800
SLC25A4	134.0	100.0	100.0	609283
SLC52A2	172.5	100.0	100.0	614707
SLC52A3	133.0	100.0	100.0	211530
SMCHD1	149.9	100.0	100.0	158901
SMDT1	125.9	100.0	100.0	-
SPEG	136.2	100.0	100.0	615959
SRPK3	98.8	100.0	99.7	-
STIM1	132.3	100.0	100.0	160565
SYT2	123.3	100.0	100.0	-
TANGO2	123.0	100.0	100.0	616878
TCAP	151.1	100.0	100.0	601954
TK2	146.7	100.0	100.0	609560
TNNI2	129.4	100.0	100.0	601680
TNNT1	122.9	100.0	100.0	605355
TNPO3	124.0	100.0	100.0	608423
TNRC6B	125.2	100.0	100.0	619243
TPM2	153.1	100.0	100.0	601680;108120;609285
TPM3	126.1	100.0	100.0	609284
TRAPPC11	147.1	100.0	100.0	615356
TRIM32	121.3	100.0	100.0	254110
TRIP4	129.4	100.0	100.0	616866
TRPV4	122.4	100.0	100.0	600175
TTC19	154.4	100.0	100.0	615157
TTN	144.9	99.6	99.1	603689;600334;608807
TUBB3	234.0	100.0	100.0	600638
TWNK	117.5	100.0	100.0	609286
UBA1	99.8	100.0	99.7	301830

UNC45B	118.5	100.0	100.0	618654;618655
VCP	126.6	100.0	100.0	167320
VIPAS39	129.6	100.0	100.0	613404
VMA21	116.7	100.0	100.0	310440
VRK1	160.5	100.0	99.8	607596
VWA1	141.6	100.0	100.0	619216
XK	96.8	100.0	99.9	300842
YARS2	139.3	100.0	100.0	613561
ZC4H2	97.2	100.0	99.9	314580

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.

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 no value for coverage are non protein coding genes.

Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.

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