

WES MUSCLE DISORDERS DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ACADVL	124.7	99.7	96.6	201475
ACTA1	83.4	98.2	89.5	255310
ACVR1	145.3	100.0	99.9	135100
AGL	153.8	99.8	99.5	232400
AGRN	151.7	97.6	92.6	254300
ANO5	133.4	99.2	96.9	611307
ATP2A1	152.6	100.0	100.0	601003
ATP7A	117.5	98.7	96.0	309400
B3GALNT2	98.9	94.3	89.8	615181
B4GAT1	120.2	100.0	100.0	615287
BAG3	167.9	100.0	99.7	612954
BICD2	133.2	99.9	99.1	615290
BIN1	115.5	99.7	96.0	255200
CACNA1S	123.6	100.0	99.8	170400
CAPN3	103.1	97.7	96.3	253600
CASQ1	101.6	99.8	96.5	616231
CAV3	222.9	100.0	100.0	614321;606072;607801
CAVIN1	154.4	100.0	100.0	613327
CCDC78	132.0	100.0	100.0	614807
CFL2	147.4	99.5	99.0	610687
CHAT	119.7	93.1	85.1	254210
CHCHD10	27.7	57.8	42.0	615048
CHKB	111.3	100.0	99.6	602541
CHRNA1	112.5	100.0	99.6	601462
CHRNB1	148.0	100.0	99.5	601462

CHRND	156.7	99.4	97.4	601462
CHRNE	156.2	100.0	100.0	601462
CHRNG	151.9	100.0	100.0	265000
CLCN1	125.3	99.9	98.8	160800;255700
CNTN1	127.3	99.7	98.6	612540
COL12A1	134.9	99.9	99.3	616471
COL13A1	86.6	93.9	93.5	616720
COL6A1	162.6	100.0	99.7	158810;254090
COL6A2	180.4	100.0	99.8	158810;254090;255600
COL6A3	156.0	100.0	99.7	158810;254090
COLQ	103.2	99.8	97.1	603034
CPT2	148.5	98.2	97.4	255110
CRPPA	105.9	98.4	94.7	614643
CRYAB	95.9	100.0	98.2	608810
DAG1	202.8	100.0	99.9	613818
DARS2	120.7	94.8	93.8	611105
DES	115.4	100.0	99.6	601419
DGUOK	112.4	99.9	98.8	617070
DMD	115.1	99.5	98.1	310200;300376
DNA2	132.9	99.6	96.9	615156
DNAJB6	61.8	95.9	84.6	611332
DNM2	133.5	98.6	93.9	160150
DOK7	123.9	94.9	92.0	254300
DPAGT1	79.4	100.0	99.8	614750
DPM1	129.7	97.4	90.9	608799
DPM2	87.3	100.0	97.7	615042
DPM3	229.1	100.0	100.0	612937
DYNC1H1	140.2	99.9	99.3	158600
DYSF	141.1	100.0	99.8	253601;254130
ECEL1	103.5	95.9	91.8	615065

EMD	137.4	99.8	97.9	310300
ENO3	173.9	100.0	100.0	131370
ERBB3	114.2	100.0	99.3	607598
EXOSC8	75.3	98.7	90.0	616081
FAM111B	146.1	99.9	99.7	615704
FHL1	64.9	99.4	93.8	300718;300719;300695;300696
FKBP14	88.0	99.8	98.7	614557
FKRP	140.6	100.0	100.0	236670;253280;606612;607155
FKTN	108.4	99.8	95.2	236670;611588;253800
FLNC	160.9	100.0	99.4	614065;609524
GAA	177.6	100.0	99.9	232300
GATM	140.6	100.0	100.0	612718
GBE1	172.1	99.9	99.7	232500
GFPT1	164.5	99.9	99.4	610542
GMPPB	257.0	100.0	100.0	615350
GNE	118.9	100.0	99.5	600737;605820
GRIN1	162.9	100.0	99.9	614254
GYG1	134.7	99.6	97.4	613507
GYS1	119.5	100.0	98.0	611556
HSPG2	123.6	99.2	97.5	255800;224410
IGHMBP2	116.6	99.3	96.9	604320
INPP5K	95.6	100.0	99.7	617404
ISCU	115.9	100.0	100.0	255125
ITGA7	141.7	99.7	97.9	613204
KBTBD13	125.9	99.9	96.7	609273
KCNJ2	149.7	100.0	100.0	170390
KIF21A	127.5	99.7	98.9	135700
KLHL40	129.7	100.0	100.0	615348
KLHL41	169.0	99.9	99.4	615731
KLHL9	194.7	100.0	99.9	-

LAMA2	131.5	99.9	99.1	607855
LAMP2	88.2	99.3	96.0	300257
LARGE1	110.3	100.0	99.7	608840
LDB3	154.8	95.4	94.8	609452
LDHA	69.1	94.4	89.3	612933
LMNA	100.3	96.1	90.6	159001;151660;613205;605588;181350
LMOD3	142.1	99.8	99.0	616165
LPIN1	123.7	99.4	97.2	605518
MAP3K20	112.4	99.9	99.2	617760
MEGF10	130.3	100.0	99.9	614399
MICU1	112.0	97.3	92.2	615673
MLIP	149.8	99.9	98.8	-
MSTN	179.4	100.0	99.9	614160
MTM1	78.0	98.7	92.0	310400
MUSK	133.1	100.0	99.9	608931
MYH2	114.3	99.9	99.1	605637
MYH3	99.6	99.9	98.4	601680
MYH7	99.2	99.1	96.7	608358;160500
MYOT	141.1	100.0	99.2	182920;159000;609200
MYPN	134.3	100.0	99.5	617336
NEB	110.3	82.9	82.5	256030
NEFH	113.7	96.3	88.2	616924
OPA1	128.7	99.5	96.7	125250
ORAI1	200.7	99.3	97.1	615883
PABPN1	70.7	64.8	54.7	164300
PFKM	114.7	100.0	99.7	232800
PGAM2	168.4	100.0	100.0	261670
PGK1	43.3	90.3	73.2	300653
PGM1	124.5	94.2	94.1	612934
PHKA1	100.8	97.8	93.4	300559

PHOX2A	36.3	92.8	74.4	602078
PIEZO2	106.7	99.8	99.2	108145;114300
PIP5K1C	142.3	99.2	96.7	611369
PLEC	130.8	100.0	99.9	613723
PNPLA2	137.8	99.8	96.1	610717
POLG	113.2	99.9	98.8	157640
POLG2	213.3	99.3	97.2	610131
POMGNT1	116.4	100.0	99.8	613157;253280;606822
POMGNT2	182.2	100.0	100.0	614830
POMK	152.4	100.0	100.0	615249
POMT1	131.1	99.5	97.3	609308;236670
POMT2	98.9	99.8	97.3	253280;613158
PREPL	108.8	98.9	96.6	616224
PRPS1	103.4	86.4	86.3	301835
PTRH2	223.0	100.0	100.0	616263
PYGM	128.2	100.0	100.0	232600
RAPSN	159.8	100.0	99.6	608931
RBCK1	102.4	99.9	98.3	615895
RRM2B	147.5	100.0	99.8	612075
RXYLT1	145.6	99.2	95.9	615041
RYR1	121.5	97.1	94.0	117000;255320
SCN4A	175.3	99.9	99.4	170500;168300;608390;613345
SELENON	137.6	84.3	84.0	255310;602771
SGCA	158.3	100.0	99.6	608099
SGCB	158.3	97.8	96.5	604286
SGCD	71.2	99.6	96.5	601287
SGCG	120.1	100.0	99.4	253700
SIL1	148.1	98.7	96.0	248800
SLC25A4	126.5	100.0	99.8	609283
SLC52A2	171.9	100.0	100.0	614707

SLC52A3	127.1	100.0	100.0	211530
SMCHD1	101.6	99.3	96.4	158901
SMDT1	155.0	100.0	100.0	-
SPEG	126.2	97.2	91.1	615959
SRPK3	123.3	99.0	95.7	-
STIM1	121.5	99.9	97.5	160565
SYT2	90.9	100.0	98.8	-
TANGO2	127.0	100.0	99.3	616878
TCAP	105.8	100.0	100.0	601954
TK2	104.4	99.0	96.0	609560
TNNI2	139.4	100.0	99.9	601680
TNNT1	108.1	99.6	97.1	605355
TNPO3	112.8	100.0	99.8	608423
TPM2	112.3	100.0	99.8	601680;108120;609285
TPM3	76.0	87.7	84.3	609284
TRAPPC11	133.5	99.7	98.7	615356
TRIM32	119.5	100.0	99.9	254110
TRIP4	105.7	99.8	99.0	616866
TRPV4	151.2	100.0	99.9	600175
TTC19	73.2	83.8	74.1	615157
TTN	170.8	98.5	98.0	603689;600334;608807
TUBB3	118.3	98.5	96.8	600638
TWNK	173.4	100.0	99.9	609286
UBA1	134.8	99.2	97.3	301830
VCP	108.5	100.0	99.1	167320
VIPAS39	123.2	100.0	100.0	613404
VMA21	82.3	99.0	92.6	310440
VRK1	133.6	99.4	97.8	607596
VWA1	76.3	85.4	77.7	619216
XK	76.5	99.7	97.6	300842

YARS2	171.1	99.9	99.4	613561
ZC4H2	78.1	100.0	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.

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.

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