WES FETAL AKINESIA DG 3.6

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACTA1	190.5	100.0	100.0	161800;255310
ADCY6	131.2	100.0	100.0	616287
ADGRG6	149.4	100.0	99.8	616503
ALG3	143.9	100.0	100.0	601110
ASCC1	117.4	86.7	86.6	616867
ATP1A2	132.9	100.0	100.0	619602
BICD2	119.7	100.0	100.0	618291
BIN1	119.6	100.0	100.0	255200
CACNA1S	121.7	100.0	100.0	No OMIM phenotype
CHAT	116.7	100.0	100.0	254210
CHRNA1	131.3	100.0	100.0	253290
CHRNB1	122.7	100.0	100.0	No OMIM phenotype
CHRND	126.6	100.0	100.0	253290
CHRNE	145.8	100.0	100.0	608931
CHRNG	123.2	100.0	100.0	265000;253290
CHST14	124.8	100.0	100.0	601776
CNTNAP1	112.2	100.0	100.0	616286
COL6A1	129.9	100.0	100.0	158810
COL6A2	134.0	100.0	100.0	158810
COL6A3	129.6	100.0	100.0	158810
COX15	126.4	100.0	100.0	615119
CRPPA	154.2	100.0	100.0	614643
DHCR24	123.9	100.0	100.0	602398
DHCR7	129.7	100.0	100.0	270400
DNM2	114.9	100.0	100.0	615368

DOK7	126.1	100.0	100.0	254300;208150
ECEL1	131.0	100.0	100.0	615065
EGR2	137.0	100.0	100.0	605253
ERBB3	123.4	100.0	100.0	607598
ERCC5	144.3	100.0	100.0	616570
ERCC6	143.9	100.0	100.0	214150
FBN2	141.3	100.0	100.0	121050
FKRP	135.4	100.0	100.0	613153
FLNC	132.3	100.0	100.0	No OMIM phenotype
FLVCR2	130.2	100.0	100.0	225790
GBA	136.2	100.0	100.0	608013
GBE1	156.7	100.0	99.9	232500
GLDN	143.4	100.0	100.0	617194
GLE1	124.9	100.0	100.0	253310;611890
GMPPB	122.5	100.0	100.0	615351
IGHMBP2	128.5	100.0	100.0	604320
KIAA1109	146.5	100.0	99.9	617822
KIF5C	137.3	99.3	99.3	615282
KLHL40	126.1	100.0	100.0	615348
KLHL41	149.1	100.0	100.0	615731
LGI4	130.6	100.0	100.0	617468
LMNA	136.0	100.0	100.0	613205
LMOD3	139.1	100.0	100.0	616165
MAGEL2	150.0	100.0	100.0	615547
MEGF10	129.9	100.0	100.0	614399
MPZ	119.2	100.0	100.0	605253
MTM1	108.2	99.7	99.2	310400
MUSK	131.5	100.0	100.0	208150
MYBPC1	136.7	100.0	100.0	614915;614335
MYCN	113.8	100.0	100.0	164280

MYH3	150.8	100.0	100.0	601680;178110;193700
MYH8	177.9	100.0	100.0	158300
MYLPF	114.0	100.0	100.0	619110
NEB	145.8	99.7	99.2	256030
NEK9	132.0	100.0	100.0	617022
NUP88	130.3	100.0	100.0	618393
PHGDH	136.5	100.0	100.0	256520
PIEZO2	133.8	100.0	100.0	617146;108145;114300
PIP5K1C	125.9	100.0	100.0	611369
PLOD1	111.4	100.0	100.0	225400
PSAT1	129.2	100.0	100.0	616038
PTRH2	156.6	100.0	100.0	616263
RAPSN	110.4	100.0	100.0	616326;208150
RIPK4	136.5	100.0	100.0	263650
RYR1	118.9	100.0	99.9	255320
SCN4A	128.7	100.0	100.0	170500;168300;608390;613345
SCYL2	154.6	100.0	100.0	618766
SELENON	108.2	93.1	91.1	255310
SLC5A7	138.4	100.0	100.0	617143
SLC6A9	124.0	100.0	100.0	617301
SMN1	127.3	93.9	93.9	253300
SMPD4	133.2	100.0	100.0	618622
STAC3	124.2	100.0	100.0	No OMIM phenotype
SYNE1	135.9	99.8	99.5	618484
TBCD	131.3	100.0	100.0	617193
TNNI2	129.4	100.0	100.0	601680
TNNT3	131.9	100.0	100.0	601680
TOR1A	132.3	91.2	90.6	618947
TPM2	153.1	100.0	100.0	601680;108120;609285
TPM3	126.1	100.0	100.0	255310

TRIP4	129.4	100.0	100.0	616866
TRPV4	122.4	100.0	100.0	600175;156530
TTN	144.9	99.6	99.1	603689;600334;608807
TUBA1A	199.9	100.0	100.0	611603
TUBB2B	285.0	100.0	100.0	610031
UBA1	99.8	100.0	99.7	301830
VIPAS39	129.6	100.0	100.0	613404
VPS33B	128.9	100.0	100.0	208085
WDR62	122.3	100.0	100.0	604317
ZC4H2	97.2	100.0	99.9	314580
ZMPSTE24	152.4	100.0	100.0	275210

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.

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