WES FETAL AKINESIA DG 3.2

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACTA1	83.4	98.2	89.5	161800;255310
ADCY6	159.7	100.0	100.0	616287
ADGRG6	145.6	99.7	98.7	616503
ALG3	90.4	100.0	99.5	601110
ASCC1	129.5	92.5	89.4	616867
BICD2	133.2	99.9	99.1	618291
BIN1	115.5	99.7	96.0	255200
CACNA1S	123.6	100.0	99.8	No OMIM phenotype
CHAT	119.7	93.1	85.1	254210
CHRNA1	112.5	100.0	99.6	253290
CHRNB1	148.0	100.0	99.5	No OMIM phenotype
CHRND	156.7	99.4	97.4	253290
CHRNE	156.2	100.0	100.0	608931
CHRNG	151.9	100.0	100.0	265000;253290
CHST14	149.1	99.9	98.8	601776
CNTNAP1	162.1	100.0	99.8	616286
COL6A1	162.6	100.0	99.7	158810
COL6A2	180.4	100.0	99.8	158810
COL6A3	156.0	100.0	99.7	158810
COX15	91.4	99.9	97.8	615119
CRPPA	105.9	98.4	94.7	614643
DHCR24	154.7	97.7	97.7	602398
DHCR7	147.5	100.0	100.0	270400
DNM2	133.5	98.6	93.9	615368
DOK7	123.9	94.9	92.0	254300;208150

ECEL1	103.5	95.9	91.8	615065
EGR2	134.7	100.0	100.0	605253
ERBB3	114.2	100.0	99.3	607598
ERCC5	118.6	99.9	99.0	616570
ERCC6	169.3	100.0	100.0	214150
FBN2	149.7	100.0	99.8	121050
FKRP	140.6	100.0	100.0	613153
FLNC	160.9	100.0	99.4	No OMIM phenotype
FLVCR2	124.9	100.0	100.0	225790
GBA	179.6	100.0	100.0	608013
GBE1	172.1	99.9	99.7	232500
GLDN	106.1	95.8	91.5	617194
GLE1	96.5	100.0	99.9	253310;611890
GMPPB	257.0	100.0	100.0	615351
IGHMBP2	116.6	99.3	96.9	604320
KIAA1109	143.1	99.8	99.0	617822
KIF5C	101.4	99.6	97.0	615282
KLHL40	129.7	100.0	100.0	615348
KLHL41	169.0	99.9	99.4	615731
LGI4	100.8	99.7	97.9	617468
LMNA	100.3	96.1	90.6	613205
LMOD3	142.1	99.8	99.0	616165
MAGEL2	114.3	94.1	89.1	615547
MEGF10	130.3	100.0	99.9	614399
MPZ	95.0	85.6	81.9	605253
MTM1	78.0	98.7	92.0	310400
MUSK	133.1	100.0	99.9	208150
MYBPC1	131.0	99.8	99.1	614915;614335
MYCN	164.2	100.0	100.0	164280
MYH3	99.6	99.9	98.4	601680;178110;193700

MYH8	120.9	100.0	99.2	158300
MYLPF	136.0	100.0	100.0	619110
NEB	110.3	82.9	82.5	256030
NEK9	116.2	99.9	99.0	617022
NUP88	140.7	99.8	99.8	618393
PHGDH	111.5	99.9	98.2	256520
PIEZO2	106.7	99.8	99.2	617146;108145;114300
PIP5K1C	142.3	99.2	96.7	611369
PLOD1	140.5	100.0	98.2	225400
PSAT1	43.1	92.0	75.1	616038
PTRH2	223.0	100.0	100.0	616263
RAPSN	159.8	100.0	99.6	616326;208150
RIPK4	151.7	100.0	99.9	263650
RYR1	121.5	97.1	94.0	255320
SCN4A	175.3	99.9	99.4	170500;168300;608390;613345
SCYL2	61.9	96.2	88.0	618766
SELENON	137.6	84.3	84.0	255310
SLC5A7	97.6	100.0	100.0	617143
SLC6A9	155.6	100.0	99.6	617301
SMN1	104.5	99.7	96.1	253300
SMPD4	98.8	99.6	95.0	618622
STAC3	118.7	100.0	100.0	No OMIM phenotype
SYNE1	127.1	98.1	97.5	618484
TBCD	136.5	95.5	93.3	617193
TNNI2	139.4	100.0	99.9	601680
TNNT3	151.5	100.0	99.6	601680
TOR1A	127.6	91.3	91.3	618947
TPM2	112.3	100.0	99.8	601680;108120;609285
TPM3	76.0	87.7	84.3	255310
TRIP4	105.7	99.8	99.0	616866

TRPV4	151.2	100.0	99.9	600175;156530
TTN	170.8	98.5	98.0	603689;600334;608807
TUBA1A	76.0	99.5	93.2	611603
TUBB2B	67.3	100.0	99.7	610031
UBA1	134.8	99.2	97.3	301830
VIPAS39	123.2	100.0	100.0	613404
VPS33B	105.9	100.0	99.9	208085
WDR62	164.4	100.0	99.9	604317
ZC4H2	78.1	100.0	98.1	314580
ZMPSTE24	134.5	99.6	99.4	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.