WES FETAL AKINESIA DG 3.4

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACTA1	249.8	100.0	100.0	161800;255310
ADCY6	193.2	100.0	100.0	616287
ADGRG6	156.1	100.0	100.0	616503
ALG3	189.5	100.0	100.0	601110
ASCC1	124.5	87.1	87.1	616867
BICD2	208.5	100.0	100.0	618291
BIN1	183.0	100.0	100.0	255200
CACNA1S	174.6	100.0	100.0	No OMIM phenotype
CHAT	171.5	100.0	100.0	254210
CHRNA1	158.2	100.0	100.0	253290
CHRNB1	180.4	100.0	100.0	No OMIM phenotype
CHRND	195.3	100.0	100.0	253290
CHRNE	187.3	100.0	100.0	608931
CHRNG	188.0	100.0	100.0	265000;253290
CHST14	217.4	100.0	100.0	601776
CNTNAP1	175.3	100.0	100.0	616286
COL6A1	211.5	100.0	100.0	158810
COL6A2	221.0	100.0	100.0	158810
COL6A3	194.7	100.0	100.0	158810
COX15	166.9	100.0	100.0	615119
CRPPA	137.3	100.0	100.0	614643
DHCR24	174.1	97.7	97.7	602398
DHCR7	189.2	100.0	100.0	270400
DNM2	179.5	100.0	100.0	615368
DOK7	223.3	100.0	100.0	254300;208150

ECEL1	192.6	100.0	100.0	615065
EGR2	236.5	100.0	100.0	605253
ERBB3	197.8	100.0	100.0	607598
ERCC5	191.7	100.0	100.0	616570
ERCC6	175.3	100.0	100.0	214150
FBN2	164.9	100.0	100.0	121050
FKRP	223.2	100.0	100.0	613153
FLNC	222.4	100.0	100.0	No OMIM phenotype
FLVCR2	197.1	100.0	100.0	225790
GBA	231.0	100.0	100.0	608013
GBE1	155.9	100.0	100.0	232500
GLDN	163.1	100.0	100.0	617194
GLE1	163.9	100.0	100.0	253310;611890
GMPPB	194.2	100.0	100.0	615351
IGHMBP2	196.2	100.0	100.0	604320
KIAA1109	159.0	100.0	100.0	617822
KIF5C	202.0	99.8	99.8	615282
KLHL40	203.3	100.0	100.0	615348
KLHL41	172.4	100.0	100.0	615731
LGI4	258.8	100.0	100.0	617468
LMNA	185.3	100.0	100.0	613205
LMOD3	200.0	100.0	100.0	616165
MAGEL2	255.3	100.0	100.0	615547
MEGF10	159.5	100.0	100.0	614399
MPZ	199.3	100.0	100.0	605253
MTM1	143.4	100.0	100.0	310400
MUSK	166.9	100.0	100.0	208150
MYBPC1	156.3	100.0	100.0	614915;614335
MYCN	198.0	100.0	100.0	164280
MYH3	198.1	100.0	100.0	601680;178110;193700

MYH8	210.3	100.0	100.0	158300
MYLPF	188.0	100.0	100.0	619110
NEB	180.5	99.9	99.9	256030
NEK9	159.3	100.0	100.0	617022
NUP88	157.2	100.0	100.0	618393
PHGDH	190.3	100.0	100.0	256520
PIEZO2	173.1	100.0	100.0	617146;108145;114300
PIP5K1C	201.5	100.0	100.0	611369
PLOD1	163.3	100.0	100.0	225400
PSAT1	169.2	100.0	100.0	616038
PTRH2	211.3	100.0	100.0	616263
RAPSN	193.2	100.0	100.0	616326;208150
RIPK4	239.3	100.0	100.0	263650
RYR1	180.0	100.0	99.9	255320
SCN4A	193.6	100.0	100.0	170500;168300;608390;613345
SCYL2	153.7	100.0	100.0	618766
SELENON	145.3	93.0	91.5	255310
SLC5A7	170.2	100.0	100.0	617143
SLC6A9	212.0	100.0	100.0	617301
SMN1	158.4	94.6	94.6	253300
SMPD4	184.3	100.0	100.0	618622
STAC3	165.6	100.0	100.0	No OMIM phenotype
SYNE1	170.2	98.8	98.8	618484
TBCD	191.3	100.0	100.0	617193
TNNI2	196.1	100.0	100.0	601680
TNNT3	181.4	100.0	100.0	601680
TOR1A	172.1	92.9	91.5	618947
TPM2	184.0	100.0	100.0	601680;108120;609285
TPM3	156.5	100.0	100.0	255310
TRIP4	150.4	100.0	100.0	616866

TRPV4	193.7	100.0	100.0	600175;156530
TTN	186.3	100.0	100.0	603689;600334;608807
TUBA1A	290.3	100.0	100.0	611603
TUBB2B	347.5	100.0	100.0	610031
UBA1	174.4	100.0	99.8	301830
VIPAS39	140.0	100.0	100.0	613404
VPS33B	151.1	100.0	100.0	208085
WDR62	197.3	100.0	100.0	604317
ZC4H2	162.0	100.0	100.0	314580
ZMPSTE24	139.3	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.