

# WES FETAL AKINESIA DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ACTA1	99.7	99.2	95.3	161800;255310
ADCY6	172.1	100.0	100.0	616287
ADGRG6	147.6	99.8	98.0	616503
ALG3	132.9	100.0	100.0	601110
ASCC1	139.1	96.7	93.2	616867
BIN1	100.2	99.1	95.3	255200
CHAT	130.3	89.3	86.8	254210
CHRNA1	121.8	94.7	94.6	253290
CHRND	150.5	100.0	99.0	253290
CHRNE	127.7	99.3	95.8	608931
CHRNA1	155.2	100.0	100.0	265000;253290
CHST14	165.6	95.7	93.3	601776
CNTNAP1	161.2	99.2	97.5	616286
COL6A1	137.2	99.5	97.8	158810
COL6A2	165.3	99.3	98.4	158810
COL6A3	174.7	100.0	99.9	158810
COX15	98.6	100.0	99.7	615119
DHCR24	183.0	100.0	100.0	602398
DHCR7	158.3	100.0	100.0	270400
DNM2	127.4	97.5	94.4	615368
DOK7	105.7	93.3	92.5	208150;254300
ECEL1	100.7	88.8	83.1	615065
EGR2	124.4	100.0	100.0	605253
ERBB3	139.2	100.0	99.9	607598
ERCC5	139.8	100.0	99.4	616570

ERCC6	191.3	100.0	99.9	214150
FBN2	161.7	100.0	99.5	121050
FKRP	94.5	100.0	99.7	613153
FLVCR2	159.7	100.0	100.0	225790
GBA	240.3	100.0	100.0	608013
GBE1	145.5	99.6	97.2	232500
GLDN	130.4	98.2	91.8	617194
GLE1	110.8	100.0	99.7	611890;253310
GMPPB	228.5	100.0	100.0	615351
IGHMBP2	107.8	99.3	96.0	604320
ISPD	104.4	95.2	84.8	614643
KIF5C	116.3	99.9	99.1	615282
KLHL40	157.9	100.0	100.0	615348
KLHL41	203.5	100.0	99.6	615731
LGI4	73.8	99.0	95.8	617468
LMNA	89.2	97.9	91.3	613205
LMOD3	141.5	99.9	98.5	616165
MEGF10	154.3	100.0	99.8	614399
MPZ	123.4	100.0	99.3	605253
MTM1	93.7	99.2	93.0	310400
MUSK	159.4	100.0	99.9	208150
MYBPC1	150.7	99.9	99.4	614915;614335
MYCN	94.1	94.9	84.8	164280
MYH3	110.4	99.9	98.6	178110;193700;601680
MYH8	134.9	100.0	99.4	158300
NEB	124.0	82.9	81.9	256030
PHGDH	115.6	100.0	99.8	256520
PIEZO2	126.1	99.9	99.2	108145;617146;114300
PIP5K1C	107.6	96.3	95.1	611369
PLOD1	137.9	99.8	97.5	225400

PSAT1	53.2	91.4	75.8	616038
RAPSN	140.5	99.6	96.3	208150;616326
RIPK4	163.3	100.0	99.6	263650
RYR1	120.7	96.8	93.7	255320
SCN4A	214.0	99.9	99.5	168300;170500;608390;613345
SELENON	111.7	85.2	83.3	255310
SLC5A7	117.1	100.0	99.9	617143
SLC6A9	161.2	100.0	99.4	617301
SMN1	112.7	99.8	96.5	253300
TBCD	152.9	95.5	92.3	617193
TNNI2	121.2	100.0	99.6	601680
TNNT3	121.0	99.9	97.8	601680
TPM2	109.1	100.0	99.6	609285;601680;108120
TPM3	98.9	89.4	89.1	255310
TRIP4	113.5	100.0	98.8	616866
TRPV4	172.4	99.5	98.7	156530;600175
TTN	187.8	98.2	97.2	608807;603689;600334
UBA1	162.0	99.8	98.9	301830
VIPAS39	144.6	100.0	100.0	613404
VPS33B	138.3	100.0	100.0	208085
WDR62	161.5	100.0	99.7	604317
ZC4H2	78.6	99.8	98.1	314580
ZMPSTE24	113.3	100.0	99.1	275210

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.

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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

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