

WES FETAL AKINESIA DG 3.1

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ACTA1	86.4	99.6	92.3	161800;255310
ADCY6	167.0	100.0	100.0	616287
ADGRG6	160.5	99.9	99.0	616503
ALG3	92.5	100.0	99.7	601110
ASCC1	151.2	93.4	90.3	616867
BICD2	137.0	100.0	99.7	618291
BIN1	119.1	99.6	95.7	255200
CACNA1S	130.8	100.0	99.9	No OMIM phenotype
CHAT	127.1	93.5	85.7	254210
CHRNA1	120.4	100.0	99.2	253290
CHRNB1	163.0	100.0	99.4	No OMIM phenotype
CHRND	158.8	99.7	97.9	253290
CHRNE	156.9	100.0	100.0	608931
CHRNA1	149.9	100.0	100.0	265000;253290
CHST14	147.4	99.9	98.9	601776
CNTNAP1	173.7	100.0	99.8	616286
COL6A1	149.2	100.0	99.4	158810
COL6A2	161.0	100.0	99.8	158810
COL6A3	171.8	100.0	99.8	158810
COX15	106.0	99.9	98.8	615119
CRPPA	122.2	98.5	94.8	614643
DHCR24	165.8	97.7	97.7	602398
DHCR7	152.1	100.0	100.0	270400
DNM2	138.9	98.1	94.5	615368
DOK7	103.6	95.1	91.6	254300;208150

ECEL1	99.8	95.4	90.0	615065
EGR2	152.7	100.0	100.0	605253
ERBB3	132.9	100.0	99.8	607598
ERCC5	143.4	100.0	99.7	616570
ERCC6	197.0	100.0	100.0	214150
FBN2	171.3	100.0	99.9	121050
FKRP	124.4	100.0	100.0	613153
FLNC	158.4	100.0	99.6	No OMIM phenotype
FLVCR2	140.7	100.0	100.0	225790
GBA	202.3	100.0	100.0	608013
GBE1	200.1	100.0	99.6	232500
GLDN	114.0	94.6	91.0	617194
GLE1	103.6	100.0	100.0	253310;611890
GMPPB	245.6	100.0	100.0	615351
IGHMBP2	118.2	98.8	95.1	604320
KIAA1109	170.5	99.8	99.2	617822
KIF5C	123.5	99.9	98.8	615282
KLHL40	134.3	100.0	100.0	615348
KLHL41	218.5	100.0	99.9	615731
LGI4	93.7	99.9	97.9	617468
LMNA	97.4	97.4	91.9	613205
LMOD3	159.8	100.0	99.7	616165
MEGF10	147.8	100.0	100.0	614399
MPZ	104.4	87.9	84.1	605253
MTM1	95.6	99.0	93.3	310400
MUSK	155.4	100.0	99.9	208150
MYBPC1	151.6	99.9	99.5	614915;614335
MYCN	140.2	100.0	99.9	164280
MYH3	113.3	99.9	99.0	601680;178110;193700
MYH8	136.9	100.0	99.6	158300

NEB	125.5	83.0	82.6	256030
NEK9	131.3	100.0	99.6	617022
NUP88	162.2	100.0	100.0	618393
PHGDH	116.1	99.9	98.8	256520
PIEZO2	121.6	100.0	99.5	617146;108145;114300
PIP5K1C	138.1	98.0	95.8	611369
PLOD1	147.6	100.0	98.4	225400
PSAT1	52.6	95.3	81.6	616038
PTRH2	259.0	100.0	100.0	616263
RAPSN	162.4	100.0	99.7	616326;208150
RIPK4	144.6	100.0	99.9	263650
RYR1	126.2	96.9	93.9	255320
SCN4A	187.3	100.0	99.6	170500;168300;608390;613345
SELENON	141.2	84.5	84.0	255310
SLC5A7	118.9	100.0	99.9	617143
SLC6A9	155.4	100.0	100.0	617301
SMN1	112.1	99.5	94.7	253300
SMPD4	96.2	99.4	94.2	618622
STAC3	132.6	100.0	100.0	No OMIM phenotype
SYNE1	146.3	98.2	97.8	618484
TBCD	147.0	96.2	94.4	617193
TNNI2	130.3	100.0	99.7	601680
TNNT3	150.9	100.0	99.7	601680
TPM2	119.2	100.0	100.0	601680;108120;609285
TPM3	85.5	89.2	87.2	255310
TRIP4	124.2	100.0	99.1	616866
TRPV4	158.6	100.0	99.9	600175;156530
TTN	198.1	98.6	98.1	603689;600334;608807
TUBA1A	77.0	99.9	97.0	611603
TUBB2B	65.7	100.0	99.5	610031

UBA1	142.9	99.4	98.2	301830
VIPAS39	133.8	100.0	100.0	613404
VPS33B	128.1	100.0	100.0	208085
WDR62	157.6	100.0	99.5	604317
ZC4H2	87.8	100.0	99.0	314580
ZMPSTE24	155.1	100.0	99.9	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.

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