

## Supplement

**eTable. Genes included in next-generation sequencing panels**

GENE	TRANSCRIPT
AARS	NM_001605.2
ACTA1	NM_001100.3
ADSSL1	NM_199165.2
AGA	NM_000027.3
AIFM1	NM_004208.3
ALS2	NM_020919.3
AMPD1	NM_000036.2
ANG	NM_001145.4
ANO5	NM_213599.2
ANXA11	NM_001157.2
APOA1	NM_000039.2
APP	NM_000484.3
ARHGEF10	NM_014629.3
ARSA	NM_000487.5
ARSB	NM_000046.3
ASAH1	NM_177924.3
ATL1	NM_015915.4
ATL3	NM_015459.4
ATP1A1	NM_000701.7
ATP2A1	NM_173201.3
ATP7A	NM_000052.6
BAG3	NM_004281.3
BICD2	NM_001003800.1

BIN1	NM_139343.2
BSCL2	NM_032667.6
CACNA1S	NM_000069.2
CASQ1	NM_001231.4
CAV3	NM_033337.2
CCDC78	NM_001031737.2
CCT5	NM_012073.4
CFL2	NM_021914.7
CHCHD10	NM_213720.2
CHMP2B	NM_014043.3
CLCN1	NM_000083.2
CLN3	NM_001042432.1
CLN5	NM_006493.2
CLN6	NM_017882.2
CLN8	NM_018941.3
CNTN1	NM_001843.3
COL12A1	NM_004370.5
COL6A1	NM_001848.2
COL6A2	NM_001849.3
COL6A3	NM_004369.3
COX6A1	NM_004373.3
CPT2	NM_000098.2
CRYAB	NM_001885.2
CTNS	NM_004937.2
CTSA	NM_000308.3
CTSD	NM_001909.4
CTSK	NM_000396.3

CYP27A1	NM_000784.3
CYP7B1	NM_004820.3
DCTN1	NM_004082.4
DES	NM_001927.3
DHTKD1	NM_018706.6
DNAJB2	NM_001039550.1
DNAJB6	NM_058246.3
DNM2	NM_001005360.2
DNMT1	NM_001130823.1
DRP2	NM_001939.2
DST	NM_001723.5;NM_015548.4
DYNC1H1	NM_001376.4
DYSF	NM_003494.3
EGR2	NM_000399.3
ELP1	NM_003640.3
ERBB4	NM_005235.2
EXOSC9	NM_001034194.1
FBLN5	NM_006329.3
FBXO38	NM_030793.4
FGD4	NM_139241.3
FHL1	NM_001449.4
FIG4	NM_014845.5
FKBP14	NM_017946.3
FLNC	NM_001458.4
FUCA1	NM_000147.4
FUS	NM_004960.3
GAA	NM_000152.3

GALC	NM_000153.3
GALNS	NM_000512.4
GAN	NM_022041.3
GARS	NM_002047.2
GDAP1	NM_018972.2
GJB1	NM_000166.5
GLA	NM_000169.2
GLB1	NM_000404.2
GM2A	NM_000405.4
GNB4	NM_021629.3
GNE	NM_001128227.2
GNPTAB	NM_024312.4
GNPTG	NM_032520.4
GNS	NM_002076.3
GRN	NM_002087.3
GSN	NM_000177.4
GUSB	NM_000181.3
GYG1	NM_004130.3
GYS1	NM_002103.4
HACD1	NM_014241.3
HARS	NM_002109.5
HEXA	NM_000520.4
HEXB	NM_000521.3
HGSNAT	NM_152419.2
HINT1	NM_005340.6
HMBS	NM_000190.3
HNRNPA2B1	NM_031243.2

HSPB1	NM_001540.3
HSPB3	NM_006308.2
HSPB8	NM_014365.2
HYAL1	NM_153281.1
IDS	NM_000202.6
IDUA	NM_000203.4
IGHMBP2	NM_002180.2
INF2	NM_022489.3
ISCU	NM_213595.3
ITM2B	NM_021999.4
KBTBD13	NM_001101362.2
KCNJ2	NM_000891.2
KCTD7	NM_153033.4
KIF1A	NM_004321.6
KIF5A	NM_004984.2
KLHL40	NM_152393.3
KLHL41	NM_006063.2
KLHL9	NM_018847.3
LAMP2	NM_002294.2
LAS1L	NM_031206.4
LDB3	NM_001080116.1;NM_001171610.1;NM_007078.3
LIPA	NM_000235.3
LITAF	NM_004862.3
LMNA	NM_170707.3
LMOD3	NM_198271.4
LRSAM1	NM_138361.5
MAN2B1	NM_000528.3

MANBA	NM_005908.3
MAP3K20	NM_016653.2
MAPT	NM_005910.5
MARS	NM_004990.3
MATR3	NM_199189.2
MCM3AP	NM_003906.4
MCOLN1	NM_020533.2
MED25	NM_030973.3
MEGF10	NM_032446.2
MFN2	NM_014874.3
MFSD8	NM_152778.2
MICAL1	NM_001286613.1
MICU1	NM_006077.3
MME	NM_007289.2
MORC2	NM_001303256.2
MPZ	NM_000530.6
MTM1	NM_000252.2
MTMR14	NM_022485.4
MTMR2	NM_016156.5
MYH2	NM_017534.5
MYH7	NM_000257.3
MYL2	NM_000432.3
MYO18B	NM_032608.6
MYOT	NM_006790.2
MYPN	NM_032578.3
NAGA	NM_000262.2
NAGLU	NM_000263.3

NDRG1	NM_006096.3
NEB	NM_001271208.1
NEFH	NM_021076.3
NEFL	NM_006158.4
NEU1	NM_000434.3
NGF	NM_002506.2
NPC1	NM_000271.4
NPC2	NM_006432.3
NTRK1	NM_001012331.1
OPTN	NM_021980.4
ORAI1	NM_032790.3
PDK3	NM_001142386.2
PFN1	NM_005022.3
PLEKHG5	NM_020631.4
PMP2	NM_002677.3
PMP22	NM_000304.3
POLG	NM_002693.2
POLG2	NM_007215.3
PPT1	NM_000310.3
PRDM12	NM_021619.2
PRNP	NM_000311.3
PRPS1	NM_002764.3
PRX	NM_181882.2
PSAP	NM_002778.3
PSEN1	NM_000021.3
PSEN2	NM_000447.2
PYROXD1	NM_024854.3

RAB7A	NM_004637.5
REEP1	NM_022912.2
RETREG1	NM_001034850.2
RYR1	NM_000540.2
SBF1	NM_002972.3
SBF2	NM_030962.3
SCN10A	NM_006514.3
SCN11A	NM_014139.2
SCN4A	NM_000334.4
SCN9A	NM_002977.3
SELENON	NM_020451.2
SEPT9	NM_006640.4
SETX	NM_015046.5
SGPL1	NM_003901.3
SGSH	NM_000199.3
SH3TC2	NM_024577.3
SIGMAR1	NM_005866.3
SLC12A6	NM_133647.1
SLC17A5	NM_012434.4
SLC25A21	NM_030631.3
SLC25A46	NM_138773.2
SLC52A1	NM_017986.3
SLC52A2	NM_024531.4
SLC52A3	NM_033409.3
SLC5A7	NM_021815.2
SMN1	NM_000344.3
SMN2	NM_017411.3



SMPD1	NM_000543.4
SNCA	NM_000345.3
SOD1	NM_000454.4
SORL1	NM_003105.5
SPEG	NM_005876.4
SPG11	NM_025137.3
SPTLC1	NM_006415.3
SPTLC2	NM_004863.3
SQSTM	NM_003900.4
STAC3	NM_145064.2
STIM1	NM_003156.3
SUMF1	NM_182760.3
SURF1	NM_003172.3
TARDBP	NM_007375.3
TAZ	NM_000116.4
TBK1	NM_013254.3
TFG	NM_006070.5
TIA1	NM_022173.2
TK2	NM_004614.4
TNNT1	NM_003283.5
TPM2	NM_003289.3
TPM3	NM_152263.3
TPP1	NM_000391.3
TREM2	NM_018965.3;NM_001271821.1
TRIM2	NM_001130067.1
TRPV4	NM_021625.4
TTN	NM_001267550.2

TTR	NM_000371.3
UBA1	NM_003334.3
UBQLN2	NM_013444.3
VAPB	NM_004738.4
VCP	NM_007126.3
VMA21	NM_001017980.3
VRK1	NM_003384.2
WNK1	NM_213655.4
YARS	NM_003680.3

All targeted regions were sequenced with  $\geq 50x$  depth, with average coverage depth of at least 350x, and that analysis focused on coding sequences, 20bp of flanking intronic regions and other specific genomic regions causative of disease at the time of assay design, as described in the genetic test results report