

WES EPILEPSY DG 3.5

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AARS1	118.3	100.0	100.0	613287;616339
ABAT	131.7	100.0	100.0	613163
ABCC8	133.6	100.0	100.0	240800
ACTB	238.4	100.0	100.0	243310
ACTL6B	115.8	100.0	100.0	618468
ACY1	118.5	100.0	100.0	609924
ADSL	139.1	100.0	100.0	103050
AGA	151.1	100.0	100.0	208400
ALDH5A1	146.0	100.0	100.0	271980
ALDH7A1	131.5	100.0	100.0	266100
ALG1	142.1	100.0	100.0	608540
ALG11	146.9	96.0	96.0	613661
ALG13	112.7	99.7	99.0	300884
ALG3	143.9	100.0	100.0	601110
ALG6	163.3	100.0	100.0	603147
AMACR	152.1	100.0	100.0	614307
AMPD2	126.3	100.0	100.0	615809
AMT	136.5	100.0	100.0	605899
ANKRD11	122.6	100.0	100.0	148050
AP1G1	140.6	100.0	100.0	619467
AP3B2	137.8	100.0	100.0	617276
ARHGEF9	95.9	96.7	95.8	300607
ARID1B	128.3	98.6	98.3	135900
ARX	109.5	99.0	96.7	308350
ASAH1	160.6	100.0	100.0	159950

ASL	121.2	100.0	100.0	207900
ASNS	154.3	100.0	100.0	615574
ASXL3	140.3	100.0	100.0	615485
ATP1A1	129.9	100.0	100.0	618314
ATP1A2	132.9	100.0	100.0	104290
ATP1A3	138.8	100.0	100.0	614820
ATP6AP2	107.6	100.0	100.0	300423
ATP7A	107.5	100.0	100.0	309400
ATRX	115.5	99.9	99.7	301042
AUTS2	135.7	100.0	100.0	615834
BOLA3	147.3	100.0	100.0	614299
BRAT1	133.4	100.0	100.0	614498
BTD	121.2	94.4	94.3	253260
CACNA1A	141.5	100.0	100.0	141500
CACNA1E	131.6	100.0	100.0	618285
CACNA2D2	126.0	100.0	100.0	618501
CACNB4	143.6	100.0	100.0	607682
CAD	129.1	100.0	100.0	616457
CASK	113.3	100.0	100.0	300749
CASQ2	141.9	100.0	100.0	611938
CCM2	113.2	100.0	100.0	603284
CDKL5	97.2	95.7	95.3	300672
CERT1	153.7	100.0	100.0	616351
CHD2	135.4	100.0	100.0	615369
CHD5	125.0	100.0	100.0	No OMIM phenotype
CHRNA2	148.8	100.0	100.0	610353
CHRNA4	131.3	100.0	100.0	600513
CHRNB2	142.1	100.0	100.0	605375
CIC	133.6	100.0	100.0	617600
CLCN4	97.0	100.0	100.0	300114

CLDN16	145.2	100.0	100.0	248250
CLDN19	139.3	100.0	100.0	248190
CLN3	121.1	93.2	93.1	204200
CLN5	135.3	83.1	83.0	256731
CLN6	141.1	100.0	100.0	601780
CLN8	131.1	100.0	100.0	610003
CNNM2	135.1	100.0	100.0	613882
CNTN2	125.4	100.0	100.0	615400
CNTNAP2	135.3	100.0	100.0	610042
COA8	146.3	100.0	99.9	220110
COL4A1	136.3	100.0	100.0	175780
COLGALT1	110.1	100.0	100.0	618360
COQ2	141.3	96.3	96.3	607426
COQ4	143.7	100.0	100.0	616276
COQ8A	127.3	100.0	100.0	612016
CPA6	150.3	100.0	100.0	614417
CPS1	146.7	100.0	100.0	237300
CPT2	139.5	100.0	100.0	600649
CSNK2B	124.2	100.0	100.0	618732
CSTB	129.2	100.0	100.0	254800
CTSD	143.8	100.0	100.0	610127
CTSF	114.0	100.0	100.0	615362
CUL4B	112.3	100.0	99.9	300354
CUX2	121.7	100.0	100.0	618141
D2HGDH	141.0	100.0	100.0	600721
DARS1	151.9	100.0	100.0	615281
DARS2	133.3	100.0	100.0	611105
DCX	109.1	98.9	98.8	300067
DDX3X	113.9	99.1	98.3	300958
DENND5A	128.3	100.0	100.0	617281

DEPDC5	134.6	100.0	100.0	604364
DHDDS	129.5	94.4	94.4	617836
DIAPH1	135.6	100.0	100.0	616632
DLAT	134.4	100.0	100.0	245348
DNAJC5	140.2	100.0	100.0	162350
DNM1	141.5	100.0	100.0	616346
DNM1L	142.0	100.0	100.0	614388
DOCK7	146.5	100.0	100.0	615730
DPAGT1	139.6	100.0	100.0	608093
DPM1	143.5	99.2	96.6	608799
DPM2	101.3	100.0	100.0	615042
DPYD	148.2	99.8	99.6	274270
DPYS	140.5	100.0	100.0	222748
DTYMK	132.0	100.0	100.0	No OMIM phenotype
DYNC1H1	125.6	100.0	100.0	614563
DYRK1A	147.1	100.0	100.0	614104
EBP	99.4	100.0	100.0	300960
EEF1A2	135.0	100.0	100.0	616409
EFHC1	135.7	97.8	97.5	254770;607631
EGF	144.0	100.0	100.0	611718
EHMT1	130.8	100.0	99.9	610253
EIF2B1	136.0	100.0	100.0	603896
EIF2B2	127.5	100.0	100.0	603896
EIF2B3	127.9	100.0	100.0	603896
EIF2B4	138.2	100.0	100.0	603896
EIF2B5	132.6	100.0	100.0	603896
EPM2A	142.9	100.0	100.0	254780
ETHE1	149.0	100.0	100.0	602473
EXOC7	121.8	100.0	100.0	619072
EXOSC3	131.1	100.0	100.0	614678

FA2H	135.3	100.0	100.0	612319
FARS2	134.7	100.0	100.0	614946
FGD1	101.6	99.9	99.5	305400
FGF12	138.2	100.0	100.0	617166
FLNA	105.8	100.0	99.9	300049
FOLR1	142.6	100.0	100.0	613068
FOXG1	123.9	100.0	99.9	613454
FOXRED1	129.6	100.0	100.0	252010
FRMPD4	92.7	100.0	99.8	300983
FRRS1L	133.9	100.0	100.0	616981
FXYD2	123.3	100.0	100.0	154020
GABRA1	158.7	100.0	100.0	615744
GABRA3	108.7	100.0	99.9	No OMIM phenotype
GABRB3	139.0	100.0	100.0	617113
GABRG2	145.5	92.9	92.6	611277
GAMT	127.9	100.0	100.0	612736
GCK	133.1	100.0	100.0	602485
GCSH	246.8	100.0	100.0	605899
GLDC	133.0	100.0	100.0	605899
GLRA1	135.9	100.0	100.0	149400
GLRB	155.4	100.0	100.0	614619
GLUD1	149.7	100.0	100.0	606762
GNAO1	124.2	100.0	100.0	615473
GOSR2	132.6	100.0	100.0	614018
GPC3	107.5	99.6	98.9	312870
GPHN	146.6	100.0	99.9	615501
GRIA3	104.8	99.7	99.2	300699
GRIN1	126.3	100.0	100.0	614254
GRIN2A	123.3	99.8	99.3	245570
GRIN2B	126.3	99.9	99.8	616139;613970

GRIN2D	110.6	99.7	98.7	617162
GRN	136.9	100.0	100.0	614706
HACE1	153.1	100.0	100.0	616756
HADH	139.9	100.0	100.0	609975
HCFC1	102.3	100.0	99.9	309541
HCN1	142.0	99.9	99.7	615871
HECW2	130.8	100.0	100.0	617268
HLCS	126.9	100.0	100.0	253270
HNRNPU	154.2	100.0	100.0	617391
HSD17B10	97.6	100.0	99.8	300438
HSD17B4	142.8	96.6	96.6	261515
IDH2	129.7	100.0	100.0	613657
IER3IP1	143.3	100.0	100.0	614231
IFIH1	150.7	100.0	100.0	615846
IQSEC2	90.8	99.7	98.4	309530
IRF2BPL	138.5	100.0	100.0	618088
ITPA	123.6	100.0	100.0	616647
JAM3	133.7	100.0	100.0	613730
KANSL1	127.7	100.0	100.0	610443
KATNB1	136.3	100.0	100.0	616212
KCNA1	140.8	100.0	100.0	160120
KCNA2	134.9	100.0	100.0	616366
KCNB1	124.9	100.0	100.0	616056
KCNC1	156.8	100.0	100.0	616187
KCNH1	124.4	98.5	98.5	611816
KCNJ10	131.0	100.0	100.0	612780
KCNJ11	127.0	100.0	100.0	601820
KCNMA1	140.0	100.0	99.9	609446
KCNQ2	137.5	100.0	100.0	613720
KCNQ3	134.7	100.0	100.0	121201

KCNT1	126.4	100.0	100.0	614959
KCNT2	151.9	99.7	99.4	617771
KCTD7	145.5	100.0	100.0	611726
KDM5C	105.5	100.0	99.9	300534
KDM6B	121.4	100.0	100.0	618505
KIF5A	120.8	100.0	100.0	617235
KMT5B	145.8	100.0	100.0	617788
KPTN	113.6	100.0	100.0	615637
KRIT1	149.8	100.0	100.0	116860
LAMB1	130.1	100.0	100.0	615191
LGI1	162.5	100.0	100.0	600512
LIAS	149.0	100.0	100.0	614462
LIPT2	172.0	100.0	100.0	617668
MAPK8IP3	138.7	100.0	100.0	618443
MBD5	139.6	100.0	100.0	156200
MECP2	88.9	100.0	99.7	312750
MED12	99.8	100.0	99.8	309520
MEF2C	148.8	100.0	100.0	613443
MFF	138.1	100.0	100.0	617086
MFSD8	156.8	100.0	100.0	610951
MLC1	126.8	100.0	100.0	604004
MOCS1	124.3	100.0	100.0	252150
MOCS2	155.4	100.0	100.0	252160
MPDU1	124.3	100.0	100.0	609180
MPDZ	141.7	99.5	99.1	615219
MTFMT	137.1	100.0	100.0	614947
MTHFR	123.4	100.0	100.0	236250
MTOR	118.8	100.0	100.0	616638
MTRR	139.1	100.0	100.0	236270
NACC1	118.7	100.0	100.0	617393

NANS	129.8	100.0	100.0	610442
NARS2	139.9	100.0	100.0	616239
NBEA	147.0	99.7	99.2	-
NCDN	123.1	100.0	100.0	619373
NDUFA1	99.1	100.0	100.0	252010
NDUFA11	112.3	100.0	98.8	252010
NDUFAF1	129.3	100.0	100.0	252010
NDUFAF2	173.6	100.0	100.0	252010
NDUFAF3	136.6	100.0	100.0	252010
NDUFAF4	154.7	100.0	100.0	252010
NDUFAF5	150.1	100.0	100.0	252010
NDUFB3	126.6	100.0	100.0	252010
NDUFB9	124.1	100.0	100.0	252010
NDUFS1	142.8	100.0	100.0	252010
NDUFS2	130.1	100.0	100.0	252010
NDUFS3	150.6	96.6	91.3	252010
NDUFS4	147.5	100.0	99.9	252010
NDUFS6	148.7	100.0	100.0	252010
NDUFV1	123.6	100.0	100.0	252010
NDUFV2	166.3	100.0	100.0	252010
NECAP1	146.1	100.0	100.0	615833
NEDD4L	139.4	100.0	100.0	617201
NEU1	118.4	100.0	100.0	256550
NEXMIF	108.2	100.0	99.9	300912
NGLY1	146.9	100.0	100.0	615273
NHLRC1	124.1	100.0	100.0	254780
NPRL2	127.5	100.0	100.0	617116
NPRL3	128.2	100.0	100.0	617118
NR2F1	139.0	100.0	99.9	615722
NRXN1	150.4	99.8	99.7	614325

NUBPL	152.9	100.0	100.0	252010
NUS1	154.9	100.0	100.0	617831
OCLN	129.7	100.0	100.0	251290
OFD1	111.2	100.0	100.0	311200
OPHN1	106.5	100.0	99.9	300486
PACS1	124.6	100.0	100.0	615009
PACS2	128.5	100.0	100.0	618067
PAFAH1B1	149.9	100.0	100.0	607432
PAK3	114.7	99.8	99.3	300558
PC	122.9	100.0	100.0	266150
PCDH19	92.4	100.0	99.9	300088
PDCD10	166.9	100.0	100.0	603285
PDHA1	104.6	99.7	97.5	312170
PDHB	128.1	100.0	100.0	614111
PDHX	138.3	100.0	99.8	245349
PDP1	139.8	100.0	100.0	608782
PDX1	144.8	100.0	100.0	245349
PET100	154.9	100.0	100.0	220110
PEX1	145.2	100.0	100.0	214100
PEX10	124.7	100.0	100.0	614870
PEX12	132.3	100.0	100.0	614859
PEX13	151.6	100.0	100.0	614883
PEX14	128.8	100.0	100.0	614887
PEX16	133.1	100.0	100.0	614876
PEX19	128.7	100.0	100.0	614886
PEX26	126.8	100.0	100.0	614872
PEX3	153.8	100.0	100.0	614882
PEX5	132.0	100.0	100.0	600414
PEX6	133.9	100.0	100.0	614862
PGAP3	122.9	100.0	100.0	615716

PHF6	122.8	100.0	100.0	301900
PHGDH	136.5	100.0	100.0	601815
PIGA	108.6	100.0	100.0	300868
PIGN	152.5	100.0	99.9	614080
PIGO	123.5	100.0	100.0	614749
PIGP	149.9	100.0	100.0	617599
PIGT	129.7	100.0	100.0	615398
PLA2G6	126.8	100.0	99.9	256600
PLCB1	150.7	100.0	100.0	613722
PLP1	95.9	99.9	98.9	312080
PLPBP	125.9	100.0	100.0	617290
PMM2	145.7	100.0	100.0	212065
PNKP	120.5	100.0	100.0	613402
PNPO	124.7	100.0	100.0	610090
POLG	138.3	100.0	100.0	203700
PPP2R1A	124.1	93.7	93.6	616362
PPP2R5D	126.6	100.0	100.0	616355
PPT1	138.1	90.3	90.3	256730
PQBP1	97.4	100.0	100.0	309500
PRF1	136.5	100.0	100.0	603553
PRICKLE1	125.3	100.0	100.0	612437
PRRT2	120.6	100.0	100.0	605751
PSAP	136.2	100.0	100.0	611722
PTRH2	156.6	100.0	100.0	616263
PTS	155.2	100.0	100.0	261640
PUM1	124.1	100.0	100.0	617931
PURA	151.7	100.0	100.0	616158
PYCR2	137.4	100.0	100.0	616420
QARS1	134.5	100.0	100.0	615760
QDPR	140.2	100.0	100.0	261630

RAB39B	123.8	100.0	100.0	300271
RARS2	144.1	100.0	100.0	611523
RNASEH2A	131.4	100.0	100.0	610333
RNASEH2B	146.8	91.4	91.4	610181
RNASEH2C	147.6	100.0	100.0	610329
ROGDI	133.7	100.0	100.0	226750
RPS6KA3	107.9	99.9	99.5	303600
RRM2B	144.5	100.0	100.0	612075
SAMHD1	149.8	100.0	100.0	612952
SCARB2	150.1	100.0	100.0	254900
SCN1A	165.6	100.0	100.0	607208
SCN1B	110.6	100.0	100.0	604233
SCN2A	167.7	100.0	100.0	613721
SCN3A	160.9	100.0	100.0	617935;617938
SCN8A	131.2	100.0	100.0	614558
SEMA6B	139.1	100.0	100.0	-
SEPSECS	141.0	100.0	100.0	613811
SERPINI1	161.9	100.0	100.0	604218
SHANK3	132.8	99.8	99.3	606232
SIK1	237.5	100.0	100.0	616341
SLC12A5	117.9	100.0	100.0	616645
SLC13A5	131.9	100.0	100.0	608305
SLC16A1	135.6	100.0	100.0	610021
SLC19A3	137.6	99.6	98.4	607483
SLC1A2	136.4	100.0	99.8	617105
SLC25A1	153.8	100.0	100.0	615182
SLC25A15	132.1	100.0	100.0	238970
SLC25A22	141.8	100.0	100.0	609304
SLC2A1	126.1	100.0	100.0	606777
SLC35A2	110.1	100.0	100.0	300896

SLC6A1	124.2	100.0	100.0	616421
SLC6A8	104.9	100.0	99.6	300352
SLC9A6	111.7	100.0	99.9	300243
SMARCA2	139.2	100.0	99.8	601358
SMC1A	96.9	100.0	99.8	300590
SMPD4	133.2	100.0	100.0	618622
SMS	103.0	100.0	99.4	309583
SNAP25	150.8	100.0	100.0	-
SPATA5	141.1	100.0	100.0	616577
SPTAN1	120.8	100.0	100.0	613477
ST3GAL3	126.9	97.4	95.3	615006
ST3GAL5	127.4	98.3	98.3	609056
STRADA	119.0	100.0	100.0	611087
STX1B	127.7	100.0	100.0	616172
STXBP1	126.8	100.0	100.0	612164
SUOX	130.2	100.0	100.0	272300
SYN1	126.2	100.0	100.0	300491
SYNGAP1	120.3	100.0	100.0	612621
SYNJ1	144.0	100.0	100.0	617389
SYP	98.7	100.0	99.8	300802
SZT2	123.3	100.0	100.0	615476
TANGO2	123.0	100.0	100.0	616878
TBC1D23	147.2	100.0	100.0	617695
TBC1D24	137.9	100.0	100.0	615338
TBCD	131.3	100.0	100.0	617193
TBCE	141.5	100.0	100.0	241410
TCF4	144.0	100.0	100.0	610954
TDP2	146.1	100.0	100.0	616949
TOE1	117.1	100.0	100.0	614969
TPP1	133.0	100.0	100.0	204500

TREX1	142.6	100.0	100.0	225750
TRIT1	130.8	100.0	100.0	617873
TRPM3	134.1	100.0	100.0	-
TRPM6	141.1	100.0	100.0	602014
TSC1	125.8	100.0	100.0	607341;191100
TSC2	134.8	100.0	100.0	613254
TSEN15	156.6	100.0	100.0	617026
TSEN2	136.8	100.0	100.0	612389
TSEN54	126.3	100.0	100.0	277470
TUBA1A	199.9	100.0	100.0	611603
TUBB2A	282.8	100.0	100.0	615763
TUBB2B	285.0	100.0	100.0	610031
TUBB4A	255.0	98.9	95.9	612438
TUBG1	157.4	100.0	100.0	615412
UBA5	153.9	100.0	100.0	617132
UBE3A	168.0	100.0	100.0	105830
UBTF	125.9	100.0	100.0	617672
UGP2	140.9	95.8	94.3	618744
VPS11	120.2	100.0	100.0	616683
VPS53	126.4	100.0	100.0	615851
WDR26	136.2	100.0	100.0	617616
WDR45	105.6	100.0	100.0	300894
WFS1	129.4	100.0	100.0	222300
WWOX	134.1	100.0	100.0	616211
XK	96.8	100.0	99.9	300842
YWHAG	130.3	100.0	100.0	617665
ZEB2	131.4	96.8	96.7	235730

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. 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.

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