

WES EPILEPSY DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AARS1	107.3	100.0	99.7	613287;616339
ABAT	92.7	99.9	97.8	613163
ABCC8	124.9	100.0	99.5	240800
ACTB	80.2	99.9	97.2	243310
ACTL6B	142.5	100.0	100.0	618468
ACY1	134.7	100.0	99.7	609924
ADSL	133.4	99.2	98.6	103050
AGA	159.9	100.0	99.9	208400
ALDH5A1	93.1	92.4	83.5	271980
ALDH7A1	65.7	91.1	84.5	266100
ALG1	44.0	53.6	46.9	608540
ALG11	133.4	96.8	96.8	613661
ALG13	81.5	97.4	90.0	300884
ALG3	90.4	100.0	99.5	601110
ALG6	111.2	98.2	93.7	603147
AMACR	151.6	100.0	100.0	614307
AMPD2	140.3	99.8	99.0	615809
AMT	158.6	100.0	100.0	605899
ANKRD11	118.2	97.0	94.0	148050
AP3B2	113.6	93.3	89.8	617276
ARHGEF9	50.0	76.3	72.5	300607
ARID1B	138.9	96.2	94.6	135900
ARX	41.3	82.1	67.5	308350
ASAH1	136.2	99.1	97.3	159950
ASL	118.5	100.0	99.7	207900

ASNS	80.7	98.1	91.2	615574
ASXL3	133.5	99.9	99.6	615485
ATP1A2	171.1	100.0	99.8	104290
ATP1A3	174.1	100.0	99.9	614820
ATP6AP2	52.2	89.9	69.7	300423
ATP7A	117.5	98.7	96.0	309400
ATRX	91.1	98.7	95.2	301042
AUTS2	140.1	98.8	96.9	615834
BOLA3	46.4	99.0	86.7	614299
BRAT1	139.8	99.9	98.9	614498
BTD	106.1	83.0	82.9	253260
CACNA1A	85.3	93.1	88.4	141500
CACNA1E	132.2	100.0	99.8	618285
CACNA2D2	128.9	93.8	93.1	618501
CACNB4	104.3	95.8	94.4	607682
CAD	139.7	99.8	98.5	616457
CASK	88.7	97.2	93.9	300749
CASQ2	125.1	100.0	99.8	611938
CDKL5	103.6	91.4	89.0	300672
CERT1	120.0	89.8	86.5	616351
CHD2	124.9	99.3	98.6	615369
CHRNA2	179.5	100.0	100.0	610353
CHRNA4	111.7	99.0	96.5	600513
CHRN2	136.6	99.3	95.5	605375
CIC	74.9	63.4	63.3	617600
CILK1	111.2	99.6	98.0	617924
CLCN4	109.2	99.9	97.7	300114
CLDN16	130.8	100.0	100.0	248250
CLDN19	136.6	98.3	92.9	248190
CLN3	128.6	92.5	92.4	204200

CLN5	106.5	69.0	66.3	256731
CLN6	136.4	99.9	98.9	601780
CLN8	152.5	83.5	83.5	610003
CNNM2	191.2	100.0	99.9	613882
CNTN2	131.7	92.7	92.7	615400
CNTNAP2	133.4	100.0	99.5	610042
COA8	71.5	81.9	80.8	220110
COL4A1	103.1	99.0	97.0	175780
COLGALT1	176.2	94.3	90.3	618360
COQ2	83.7	97.6	96.7	607426
COQ4	92.0	91.0	89.7	616276
COQ8A	166.1	100.0	99.6	612016
CPA6	116.1	99.2	96.1	614417
CPS1	138.3	100.0	100.0	237300
CPT2	148.5	98.2	97.4	600649
CSNK2B	136.8	100.0	100.0	618732
CSTB	72.5	99.6	90.5	254800
CTSD	174.4	98.4	95.0	610127
CTSF	114.5	83.9	78.9	615362
CUL4B	79.2	96.9	88.8	300354
CUX2	135.4	99.9	99.3	618141
D2HGDH	142.4	99.7	98.2	600721
DARS1	134.5	99.5	99.3	615281
DARS2	120.7	94.8	93.8	611105
DCX	97.1	100.0	98.9	300067
DDX3X	66.1	81.1	78.6	300958
DENND5A	102.7	99.8	98.7	617281
DEPDC5	130.4	99.9	99.7	604364
DHDDS	80.8	99.4	95.6	617836
DIAPH1	100.4	99.8	98.4	616632

DLAT	98.4	99.8	99.3	245348
DNAJC5	235.2	100.0	100.0	162350
DNM1	139.8	92.7	89.0	616346
DNM1L	118.8	99.6	98.3	614388
DOCK7	124.8	99.6	98.3	615730
DPAGT1	79.4	100.0	99.8	608093
DPM1	129.7	97.4	90.9	608799
DPM2	87.3	100.0	97.7	615042
DPYD	146.1	99.5	96.5	274270
DPYS	118.4	100.0	100.0	222748
DYNC1H1	140.2	99.9	99.3	614563
DYRK1A	137.1	100.0	100.0	614104
EBP	67.1	99.5	94.3	300960
EEF1A2	184.6	100.0	100.0	616409
EFHC1	126.4	92.8	91.0	254770;607631
EGF	114.9	99.9	99.8	611718
EHMT1	128.4	94.5	93.6	610253
EIF2B1	121.8	100.0	99.8	603896
EIF2B2	119.0	100.0	98.1	603896
EIF2B3	145.4	100.0	100.0	603896
EIF2B4	125.5	100.0	99.5	603896
EIF2B5	104.1	99.8	98.5	603896
EPM2A	127.1	93.9	91.2	254780
ETHE1	85.4	99.3	93.3	602473
EXOSC3	120.9	98.1	90.5	614678
FA2H	86.5	92.4	82.6	612319
FARS2	180.1	100.0	100.0	614946
FGD1	83.8	97.2	91.2	305400
FGF12	114.8	100.0	99.1	617166
FLNA	143.7	100.0	99.9	300049

FOLR1	103.2	100.0	99.9	613068
FOXG1	144.1	87.2	79.4	613454
FOXRED1	121.6	100.0	99.6	252010
FRMPD4	107.4	97.0	94.2	300983
FRRS1L	110.8	82.5	73.7	616981
FXYD2	133.7	100.0	100.0	154020
GABRA1	165.8	100.0	100.0	615744
GABRB3	129.6	99.8	98.1	617113
GABRG2	139.3	89.9	88.4	611277
GAMT	124.7	95.0	82.7	612736
GCK	130.0	95.4	95.4	602485
GCSH	27.8	75.7	64.4	605899
GLDC	57.7	88.9	77.8	605899
GLRA1	94.7	100.0	99.8	149400
GLRB	106.2	99.1	95.3	614619
GLUD1	64.6	96.4	84.4	606762
GNAO1	157.1	93.8	93.8	615473
GOSR2	136.1	96.0	95.1	614018
GPC3	77.3	98.8	92.9	312870
GPHN	153.4	99.9	99.1	615501
GRIA3	88.1	99.5	94.0	300699
GRIN1	162.9	100.0	99.9	614254
GRIN2A	140.1	100.0	100.0	245570
GRIN2B	155.5	99.6	98.7	616139;613970
GRIN2D	74.5	83.8	69.4	617162
GRN	178.3	100.0	100.0	614706
HACE1	146.1	99.7	99.3	616756
HADH	109.8	99.2	97.7	609975
HCFC1	95.0	98.1	93.0	309541
HCN1	133.8	98.4	98.3	615871

HECW2	114.5	99.8	98.2	617268
HLCS	153.7	100.0	100.0	253270
HNRNPU	144.7	99.9	98.7	617391
HSD17B10	103.8	99.9	98.3	300438
HSD17B4	114.3	95.3	92.8	261515
IDH2	108.9	99.8	97.4	613657
IER3IP1	82.2	92.0	80.2	614231
IFIH1	127.4	99.5	97.3	615846
IQSEC2	69.3	94.6	84.0	309530
IRF2BPL	160.3	100.0	97.9	618088
ITPA	126.4	100.0	100.0	616647
JAM3	134.7	100.0	100.0	613730
KANSL1	143.1	99.8	98.2	610443
KATNB1	146.7	100.0	100.0	616212
KCNA1	144.1	100.0	100.0	160120
KCNA2	132.6	100.0	99.6	616366
KCNB1	135.0	100.0	99.7	616056
KCNC1	163.2	100.0	100.0	616187
KCNH1	153.7	98.7	98.5	611816
KCNJ10	146.8	89.2	88.5	612780
KCNJ11	174.3	100.0	100.0	601820
KCNMA1	105.6	94.0	93.0	609446
KCNQ2	117.0	91.2	89.1	613720
KCNQ3	106.7	99.8	97.2	121201
KCNT1	134.5	95.8	95.0	614959
KCNT2	105.9	99.1	96.9	617771
KCTD7	161.4	95.0	95.0	611726
KDM5C	101.9	99.7	97.7	300534
KDM6B	151.7	98.6	97.4	618505
KIF5A	123.5	100.0	99.8	617235

KMT5B	164.4	99.7	98.5	617788
KPTN	137.0	100.0	100.0	615637
LAMB1	143.0	100.0	99.6	615191
LGI1	142.2	98.3	98.1	600512
LIAS	130.5	99.8	98.9	614462
MAPK8IP3	164.3	99.4	99.0	618443
MBD5	148.0	99.9	99.8	156200
MECP2	128.3	99.8	97.5	312750
MED12	88.0	99.3	94.1	309520
MEF2C	133.8	99.7	95.7	613443
MFF	88.3	93.9	89.4	617086
MFSD8	121.3	99.6	99.4	610951
MLC1	99.5	100.0	98.8	604004
MOCS1	92.4	98.9	95.5	252150
MOCS2	143.2	99.4	99.4	252160
MPDU1	105.1	100.0	99.2	609180
MPDZ	134.4	99.8	98.5	615219
MTFMT	138.9	99.9	99.5	614947
MTHFR	111.9	97.3	95.9	236250
MTOR	114.4	99.9	98.9	616638
MTRR	129.7	99.8	98.4	236270
NACC1	160.6	100.0	100.0	617393
NANS	98.3	100.0	99.9	610442
NARS2	116.3	97.9	97.1	616239
NBEA	129.9	91.8	90.3	-
NDUFA1	185.7	99.8	99.3	252010
NDUFA11	118.1	100.0	99.8	252010
NDUFAF1	116.6	100.0	100.0	252010
NDUFAF2	49.9	91.0	77.5	252010
NDUFAF3	128.1	100.0	99.9	252010

NDUF4F4	104.6	99.6	96.9	252010
NDUF4F5	119.6	99.7	99.1	252010
NDUFB3	30.8	88.6	71.0	252010
NDUFB9	99.8	96.1	91.5	252010
NDUFS1	161.0	99.9	99.1	252010
NDUFS2	98.4	100.0	100.0	252010
NDUFS3	127.5	90.7	90.6	252010
NDUFS4	156.0	99.7	99.7	252010
NDUFS6	115.6	100.0	99.8	252010
NDUFV1	138.5	99.0	97.0	252010
NDUFV2	69.8	85.8	78.7	252010
NECAP1	109.1	100.0	100.0	615833
NEDD4L	98.2	71.9	71.7	617201
NEU1	144.9	99.3	96.1	256550
NEXMIF	129.9	99.9	99.0	300912
NGLY1	142.3	99.8	99.7	615273
NHLRC1	153.0	100.0	99.8	254780
NPRL2	147.9	100.0	100.0	617116
NPRL3	126.5	100.0	99.9	617118
NRXN1	148.5	97.5	96.9	614325
NUBPL	94.2	99.5	96.9	252010
NUS1	57.9	56.5	42.0	617831
OCLN	193.1	100.0	99.9	251290
OFD1	50.3	87.1	71.3	311200
OPHN1	88.4	99.3	96.3	300486
PACS1	110.3	99.8	99.0	615009
PACS2	164.7	99.8	97.1	618067
PAFAH1B1	89.9	93.5	84.9	607432
PAK3	78.8	98.9	93.6	300558
PC	158.4	99.7	98.0	266150

PCDH19	158.0	99.7	97.7	300088
PDHA1	85.2	98.8	95.9	312170
PDHB	111.0	99.2	96.8	614111
PDHX	143.5	99.8	99.6	245349
PDP1	129.4	100.0	100.0	608782
PDX1	61.1	94.8	82.2	245349
PET100	88.7	100.0	99.2	220110
PEX1	135.8	99.8	99.4	214100
PEX10	105.9	98.8	90.6	614870
PEX12	132.6	100.0	100.0	614859
PEX13	168.6	100.0	100.0	614883
PEX14	139.1	95.8	89.4	614887
PEX16	167.1	97.1	93.9	614876
PEX19	87.0	99.0	94.4	614886
PEX26	86.7	100.0	99.8	614872
PEX3	98.2	99.4	99.2	614882
PEX5	110.1	99.9	98.8	600414
PEX6	109.2	96.4	88.0	614862
PGAP3	66.7	62.6	58.1	615716
PHF6	61.0	96.2	84.7	301900
PHGDH	111.5	99.9	98.2	601815
PIGA	73.9	91.6	82.5	300868
PIGN	96.7	93.1	89.6	614080
PIGO	149.1	100.0	99.8	614749
PIGP	76.2	95.6	85.5	617599
PIGT	168.8	98.1	98.0	615398
PLA2G6	111.6	92.1	90.7	256600
PLCB1	136.7	99.9	99.4	613722
PLP1	117.9	99.9	97.7	312080
PLPBP	90.1	95.1	88.9	617290

PMM2	127.8	99.8	99.8	212065
PNKP	115.5	100.0	100.0	613402
PNPO	73.2	99.9	97.1	610090
POLG	113.2	99.9	98.8	203700
PPP2R1A	129.7	91.6	91.6	616362
PPP2R5D	150.8	100.0	99.8	616355
PPT1	137.3	90.3	89.9	256730
PQBP1	138.7	100.0	99.3	309500
PRF1	121.1	91.2	90.1	603553
PRICKLE1	97.2	100.0	99.9	612437
PRRT2	121.3	100.0	98.8	605751
PSAP	99.1	100.0	99.6	611722
PTRH2	223.0	100.0	100.0	616263
PTS	104.8	99.5	99.0	261640
PUM1	130.9	100.0	99.4	617931
PURA	144.6	98.6	94.6	616158
PYCR2	127.2	100.0	99.3	616420
QARS1	135.7	100.0	100.0	615760
QDPR	97.7	100.0	98.9	261630
RAB39B	96.8	100.0	99.8	300271
RARS2	104.3	99.7	98.6	611523
RNASEH2A	140.3	100.0	99.7	610333
RNASEH2B	97.2	81.0	78.2	610181
RNASEH2C	271.1	100.0	100.0	610329
ROGDI	102.4	98.6	95.2	226750
RPS6KA3	92.3	98.4	91.4	303600
RRM2B	147.5	100.0	99.8	612075
SAMHD1	137.6	98.5	97.9	612952
SCARB2	108.9	99.9	99.4	254900
SCN1A	125.3	99.7	99.1	607208

SCN1B	172.2	98.2	96.3	604233
SCN2A	142.3	99.4	97.4	613721
SCN3A	150.4	99.8	99.1	617935;617938
SCN8A	168.5	100.0	99.5	614558
SEMA6B	87.0	82.4	75.5	-
SEPSECS	154.7	99.9	99.6	613811
SERPINI1	104.8	99.8	98.9	604218
SHANK3	121.9	92.4	84.9	606232
SIK1	108.5	98.2	93.9	616341
SLC12A5	110.1	83.9	83.8	616645
SLC13A5	151.1	100.0	100.0	608305
SLC16A1	124.8	100.0	98.6	610021
SLC19A3	130.4	97.8	97.0	607483
SLC1A2	97.7	96.1	94.8	617105
SLC25A1	91.4	96.9	89.8	615182
SLC25A15	161.2	99.3	96.6	238970
SLC25A22	117.3	99.2	96.5	609304
SLC2A1	164.2	92.8	92.7	606777
SLC35A2	110.5	99.6	97.7	300896
SLC6A1	128.2	96.7	96.6	616421
SLC6A8	54.5	94.8	83.0	300352
SLC9A6	106.7	94.7	90.2	300243
SMARCA2	109.2	96.7	96.3	601358
SMC1A	89.7	99.6	97.1	300590
SMPD4	98.8	99.6	95.0	618622
SMS	66.0	87.9	72.1	309583
SNAP25	115.9	100.0	99.8	-
SPATA5	137.1	99.8	99.5	616577
SPTAN1	112.6	99.1	97.9	613477
ST3GAL3	95.7	68.8	68.2	615006

ST3GAL5	94.7	85.9	84.0	609056
STX1B	144.7	100.0	100.0	616172
STXBP1	105.8	96.8	96.2	612164
SUOX	179.3	100.0	100.0	272300
SYN1	60.6	82.0	71.6	300491
SYNGAP1	154.8	98.8	97.4	612621
SYNJ1	133.5	99.7	98.1	617389
SYP	64.3	99.9	96.2	300802
SZT2	144.1	99.6	99.3	615476
TANGO2	127.0	100.0	99.3	616878
TBC1D23	95.2	98.7	94.5	617695
TBC1D24	180.7	100.0	100.0	615338
TBCD	136.5	95.5	93.3	617193
TBCE	118.0	99.7	96.6	241410
TCF4	106.4	100.0	99.9	610954
TDP2	166.5	99.6	99.5	616949
TOE1	156.3	100.0	100.0	614969
TPP1	130.4	100.0	100.0	204500
TREX1	288.9	100.0	100.0	225750
TRPM3	113.3	99.9	99.4	-
TRPM6	129.8	99.9	99.1	602014
TSC1	118.1	99.5	98.2	607341;191100
TSC2	140.3	100.0	99.8	613254
TSEN15	63.1	78.9	77.0	617026
TSEN2	108.2	99.9	99.2	612389
TSEN54	118.6	96.7	94.8	277470
TUBA1A	76.0	99.5	93.2	611603
TUBB2A	68.3	96.9	95.7	615763
TUBB2B	67.3	100.0	99.7	610031
TUBB4A	84.5	96.0	95.6	612438

TUBG1	178.0	100.0	100.0	615412
UBA5	81.6	97.4	86.6	617132
UBE3A	84.7	98.9	94.1	105830
UBTF	116.6	99.9	99.1	617672
UGP2	122.5	98.7	98.2	618744
VPS11	117.7	94.6	92.2	616683
VPS53	115.6	91.1	89.9	615851
WDR26	80.7	89.2	84.2	617616
WDR45	71.6	98.2	92.0	300894
WWOX	116.3	100.0	99.9	616211
XK	76.5	99.7	97.6	300842
YWHAG	169.1	100.0	99.9	617665
ZEB2	137.0	99.7	98.5	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.