

WES EPILEPSY DG 2.18

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AARS	124.9	100.0	99.9	613287;616339
ABAT	105.5	100.0	99.4	613163
ABCC8	132.1	100.0	99.8	240800
ACTB	74.3	99.7	96.1	243310
ACTL6B	143.9	100.0	99.8	618468
ACY1	131.7	100.0	98.8	609924
ADSL	150.2	99.2	98.7	103050
ALDH7A1	77.2	94.4	88.8	266100
ALG1	48.4	53.0	45.8	608540
ALG11	162.5	96.8	96.8	613661
ALG13	92.1	98.4	92.6	300884
ALG3	92.5	100.0	99.7	601110
ALG6	127.1	98.6	94.8	603147
AMACR	159.5	100.0	100.0	614307
AMPD2	139.8	99.8	98.9	615809
AMT	153.3	100.0	100.0	605899
ANKRD11	119.3	97.5	94.8	148050
AP3B2	140.6	99.4	95.1	617276
APOPT1	92.5	81.9	80.7	220110
ARHGEF9	59.3	76.5	74.1	300607
ARID1B	158.5	99.5	98.6	135900
ARX	38.6	81.0	64.0	308350
ASAH1	158.2	99.7	98.6	159950
ASL	113.6	100.0	99.6	207900
ASNS	98.7	99.4	95.2	615574

ASXL3	161.8	99.9	99.7	615485
ATP1A2	183.1	100.0	100.0	104290
ATP1A3	173.5	100.0	99.9	614820
ATP6AP2	56.8	94.1	76.6	300423
ATP7A	135.5	99.7	97.5	309400
ATRX	102.2	99.4	96.3	301042
AUTS2	141.1	98.2	95.8	615834
BOLA3	50.8	99.4	90.2	614299
BRAT1	131.1	99.7	98.2	614498
BTD	155.8	100.0	99.9	253260
CACNA1A	97.0	95.1	91.9	141500
CACNA1E	142.9	100.0	99.9	618285
CACNA2D2	136.9	94.0	93.2	618501
CACNB4	122.4	95.5	94.3	607682
CAD	150.4	100.0	99.2	616457
CASK	106.8	99.6	96.8	300749
CDKL5	125.4	95.0	93.5	300672
CHD2	151.3	99.4	99.2	615369
CHRNA2	185.5	100.0	100.0	610353
CHRNA4	108.8	98.3	96.2	600513
CHRN2	137.0	99.3	96.0	605375
CIC	71.6	63.3	63.3	617600
CLCN4	117.3	99.9	98.9	300114
CLDN16	162.9	100.0	100.0	248250
CLDN19	128.4	98.5	93.1	248190
CLN3	133.0	92.5	91.8	204200
CLN5	164.8	99.4	95.5	256731
CLN6	141.3	99.9	97.1	601780
CLN8	173.9	83.5	83.5	610003
CNNM2	198.1	100.0	100.0	613882

CNTN2	134.1	92.7	92.7	615400
CNTNAP2	154.0	100.0	99.8	610042
COL4A1	115.6	98.7	97.4	175780
COL4A3BP	150.5	99.6	96.5	616351
COLGALT1	181.2	93.3	89.0	618360
COQ2	113.1	98.0	95.3	607426
COQ4	98.8	90.9	89.3	616276
COQ8A	164.9	100.0	99.5	612016
CPA6	141.4	99.6	97.5	614417
CPS1	162.9	100.0	99.9	237300
CPT2	159.1	98.2	97.8	600649
CSTB	85.0	99.6	89.8	254800
CTSD	174.3	98.4	95.0	610127
CTSF	117.3	84.0	79.3	615362
CUL4B	93.0	98.0	90.9	300354
CUX2	133.0	99.9	99.1	618141
D2HGDH	135.3	99.2	97.2	600721
DCX	114.9	100.0	99.9	300067
DDX3X	86.6	86.8	84.9	300958
DENND5A	118.6	100.0	99.4	617281
DEPDC5	147.4	100.0	99.8	604364
DHDDS	93.1	99.0	95.0	617836
DLAT	111.4	100.0	99.7	245348
DNAJC5	228.0	100.0	100.0	162350
DNM1	145.0	92.6	89.1	616346
DNM1L	139.3	99.9	98.5	614388
DOCK7	150.4	99.8	98.2	615730
DPAGT1	90.7	100.0	100.0	608093
DPM1	155.7	98.2	91.3	608799
DPM2	98.8	100.0	98.7	615042

DPYD	173.0	99.7	97.7	274270
DPYS	137.1	100.0	99.9	222748
DYNC1H1	157.5	99.9	99.4	614563
DYRK1A	158.8	100.0	100.0	614104
EEF1A2	189.2	100.0	100.0	616409
EFHC1	149.7	93.1	91.6	607631;254770
EGF	129.4	99.9	99.7	611718
EHMT1	138.6	94.5	93.7	610253
EPM2A	138.3	94.2	91.5	254780
ETHE1	96.2	99.9	97.4	602473
EXOSC3	125.9	99.5	94.9	614678
FA2H	96.4	92.0	83.1	612319
FARS2	206.7	100.0	100.0	614946
FGD1	91.2	97.3	92.8	305400
FGF12	124.7	99.9	98.1	617166
FLNA	138.5	100.0	99.9	300049
FOLR1	130.7	100.0	100.0	613068
FOXG1	144.6	88.6	82.1	613454
FOXRED1	136.0	100.0	99.9	252010
FRMPD4	122.3	99.8	98.5	300983
FRRS1L	134.6	79.7	69.1	616981
FXVD2	131.5	100.0	100.0	154020
GABRA1	197.0	100.0	100.0	615744
GABRB3	147.9	99.6	98.2	617113
GABRG2	166.9	90.8	90.2	611277
GAMT	113.1	93.1	82.7	612736
GCK	132.1	100.0	100.0	602485
GCSH	33.4	75.7	68.9	605899
GLDC	70.9	89.9	82.0	605899
GLRA1	116.2	100.0	99.8	149400

GLRB	119.9	99.2	95.1	614619
GLUD1	72.8	94.2	82.9	606762
GNAO1	173.0	93.8	93.8	615473
GOSR2	149.4	95.9	94.6	614018
GPC3	90.4	99.1	94.7	312870
GPHN	175.6	100.0	99.5	615501
GRIA3	101.2	99.7	96.1	300699
GRIN1	164.3	100.0	100.0	614254
GRIN2A	161.2	100.0	100.0	245570
GRIN2B	174.2	99.8	99.2	613970;616139
GRIN2D	76.2	79.8	65.4	617162
GRN	166.2	100.0	100.0	614706
HACE1	171.9	100.0	99.3	616756
HADH	127.1	99.0	97.5	609975
HCFC1	97.0	98.3	93.6	309541
HCN1	153.5	100.0	99.7	615871
HLCS	179.3	100.0	100.0	253270
HNRNPU	170.5	99.9	98.9	617391
HSD17B10	109.8	100.0	99.1	300438
HSD17B4	130.8	96.0	93.7	261515
ICK	133.5	99.9	98.7	617924
IDH2	111.5	99.7	97.4	613657
IER3IP1	102.4	91.9	82.6	614231
IFIH1	137.3	99.7	98.4	615846
IQSEC2	72.4	96.8	88.6	309530
IRF2BPL	160.3	99.5	95.0	618088
ITPA	149.2	100.0	100.0	616647
JAM3	149.8	100.0	99.9	613730
KANSL1	179.7	99.9	99.2	610443
KATNB1	136.6	100.0	99.9	616212

KCNA1	166.8	100.0	99.9	160120
KCNA2	160.9	100.0	99.6	616366
KCNB1	157.0	100.0	99.6	616056
KCNC1	157.1	100.0	100.0	616187
KCNH1	173.4	98.7	98.7	611816
KCNJ10	157.5	89.3	89.0	612780
KCNJ11	177.3	100.0	100.0	601820
KCNMA1	122.3	94.4	93.6	609446
KCNQ2	113.6	91.3	89.8	613720
KCNQ3	123.9	100.0	99.4	121201
KCNT1	129.7	96.0	95.2	614959
KCTD7	163.6	95.0	95.0	611726
KDM5C	109.2	99.8	97.9	300534
KDM6B	161.3	98.8	97.9	618505
KPTN	143.1	100.0	100.0	615637
LAMB1	171.8	100.0	99.9	615191
LGI1	170.5	98.5	97.5	600512
LIAS	151.6	100.0	99.1	614462
MAPK8IP3	163.6	100.0	99.6	618443
MBD5	170.1	99.9	99.9	156200
MECP2	149.7	100.0	98.7	312750
MED12	101.2	99.8	96.7	309520
MEF2C	163.5	99.9	96.0	613443
MFF	101.3	94.3	89.9	617086
MFSD8	143.3	100.0	99.7	610951
MLC1	103.0	100.0	99.0	604004
MOCS1	94.4	99.2	95.4	252150
MOCS2	166.1	99.6	99.5	252160
MPDU1	112.0	100.0	100.0	609180
MPDZ	159.6	99.8	98.8	615219

MTHFR	117.2	97.3	96.0	236250
MTOR	131.9	100.0	99.5	616638
MTRR	156.8	100.0	99.6	236270
NACC1	153.1	100.0	99.8	617393
NANS	114.3	100.0	99.9	610442
NBEA	146.7	92.0	90.6	-
NDUFA1	196.4	99.9	99.3	252010
NDUFA11	117.9	100.0	100.0	252010
NDUFAB1	135.8	100.0	100.0	252010
NDUFAB2	63.6	95.0	83.4	252010
NDUFAB3	129.2	100.0	99.9	252010
NDUFAB4	141.6	99.8	98.2	252010
NDUFAB5	139.0	100.0	99.5	252010
NDUFB3	34.6	95.8	80.5	252010
NDUFB9	120.3	98.4	95.5	252010
NDUFS1	178.7	100.0	99.5	252010
NDUFS2	118.5	100.0	100.0	252010
NDUFS3	141.3	90.7	90.6	252010
NDUFS4	190.8	100.0	99.4	252010
NDUFS6	138.2	100.0	99.9	252010
NDUFV1	158.4	98.0	96.1	252010
NDUFV2	84.9	86.9	76.9	252010
NECAP1	124.2	100.0	100.0	615833
NEDD4L	116.7	72.0	71.5	617201
NEU1	146.4	99.7	97.7	256550
NEXMIF	153.1	100.0	99.5	300912
NGLY1	166.1	100.0	99.8	615273
NHLRC1	150.9	100.0	98.7	254780
NPRL2	157.9	100.0	100.0	617116
NPRL3	131.1	100.0	99.6	617118

NRXN1	161.3	97.4	96.9	614325
NUBPL	111.9	99.7	98.4	252010
OCLN	204.3	100.0	100.0	251290
OFD1	61.2	88.0	73.7	311200
OPHN1	104.2	99.5	97.6	300486
PACS1	116.2	98.8	96.9	615009
PACS2	155.5	99.3	96.2	618067
PAFAH1B1	102.1	94.1	87.1	607432
PAK3	95.4	99.3	95.9	300558
PC	161.8	99.8	97.3	266150
PCDH19	175.8	100.0	98.9	300088
PDHA1	102.0	99.4	97.1	312170
PDHB	132.1	99.1	97.5	614111
PDHX	159.4	99.9	99.4	245349
PDP1	151.0	100.0	100.0	608782
PDX1	59.1	93.0	82.4	245349
PET100	107.1	100.0	99.6	220110
PEX1	155.5	99.9	99.4	214100
PEX10	101.8	96.8	89.7	614870
PEX12	150.7	100.0	100.0	614859
PEX13	208.9	100.0	100.0	614883
PEX14	132.7	96.7	90.8	614887
PEX16	158.6	97.9	94.2	614876
PEX19	103.7	99.9	98.5	614886
PEX26	94.0	100.0	100.0	614872
PEX3	125.7	100.0	99.3	614882
PEX5	124.0	99.9	99.0	600414
PEX6	113.0	94.5	86.7	614862
PGAP3	70.6	63.5	59.6	615716
PHF6	68.5	97.8	88.3	301900

PHGDH	116.1	99.9	98.8	601815
PIGA	85.7	93.8	86.7	300868
PIGN	117.3	93.8	91.5	614080
PIGO	156.3	100.0	99.9	614749
PIGP	96.9	95.8	87.3	617599
PIGT	174.8	98.1	98.1	615398
PLA2G6	120.6	99.9	98.3	256600
PLCB1	160.9	100.0	99.8	613722
PLP1	141.7	100.0	99.2	312080
PLPBP	111.9	98.2	90.1	617290
PMM2	145.4	100.0	100.0	212065
PNKP	112.0	100.0	100.0	613402
PNPO	78.4	99.9	97.7	610090
POLG	111.6	100.0	99.3	203700
PPP2R1A	136.5	91.6	91.5	616362
PPP2R5D	177.6	100.0	100.0	616355
PPT1	164.4	90.3	90.3	256730
PQBP1	137.8	100.0	100.0	309500
PRF1	114.3	91.2	90.8	603553
PRICKLE1	118.6	100.0	100.0	612437
PRRT2	122.5	100.0	99.6	605751
PSAP	112.4	100.0	100.0	611722
PTS	118.8	99.9	99.1	261640
PUM1	148.2	100.0	99.9	617931
PURA	147.0	99.0	95.2	616158
PYCR2	118.2	100.0	99.1	616420
QARS	151.2	100.0	100.0	615760
QDPR	112.9	100.0	99.7	261630
RAB39B	109.8	100.0	100.0	300271
RARS2	122.7	100.0	99.8	611523

RNASEH2A	150.5	100.0	100.0	610333
RNASEH2B	126.7	96.0	92.5	610181
RNASEH2C	279.8	100.0	99.5	610329
ROGDI	103.5	98.4	95.2	226750
RPS6KA3	105.3	98.4	94.4	303600
RRM2B	163.8	100.0	99.7	612075
SAMHD1	164.2	100.0	99.6	612952
SCARB2	121.3	100.0	99.8	254900
SCN1A	143.7	99.9	99.5	607208
SCN1B	178.3	98.0	96.4	604233
SCN2A	165.0	99.6	97.6	613721
SCN3A	173.7	99.9	99.3	617938;617935
SCN8A	189.5	100.0	99.8	614558
SEMA6B	88.4	80.6	73.6	-
SEPSECS	192.2	100.0	100.0	613811
SERPINI1	119.3	99.9	99.0	604218
SHANK3	111.9	92.2	82.1	606232
SIK1	115.7	98.7	94.4	616341
SLC12A5	116.3	83.9	83.8	616645
SLC13A5	156.9	100.0	99.9	608305
SLC16A1	162.9	100.0	99.3	610021
SLC19A3	160.1	100.0	99.8	607483
SLC1A2	121.9	100.0	99.3	617105
SLC25A1	97.8	95.8	88.6	615182
SLC25A15	176.5	99.8	98.1	238970
SLC25A22	115.0	98.6	95.8	609304
SLC2A1	164.9	92.8	92.8	606777
SLC35A2	115.7	99.9	98.4	300896
SLC6A1	146.8	100.0	100.0	616421
SLC6A8	56.3	93.5	81.6	300352

SLC9A6	123.7	99.3	95.5	300243
SMARCA2	125.9	96.7	96.2	601358
SMC1A	102.0	100.0	98.7	300590
SMPD4	96.2	99.4	94.2	618622
SMS	80.6	91.5	78.5	309583
SNAP25	138.6	100.0	99.9	-
SPATA5	167.7	100.0	99.7	616577
SPTAN1	126.6	99.1	98.6	613477
ST3GAL3	151.5	100.0	99.8	615006
ST3GAL5	117.5	85.0	84.2	609056
STX1B	163.2	100.0	100.0	616172
STXBP1	120.1	96.8	96.5	612164
SUOX	189.7	100.0	100.0	272300
SYN1	69.2	81.9	73.2	300491
SYNGAP1	167.5	99.4	98.1	612621
SYNJ1	157.8	99.9	99.4	617389
SYP	72.5	99.9	96.7	300802
SZT2	153.3	99.6	99.5	615476
TANGO2	133.1	100.0	99.3	616878
TBC1D23	113.4	99.7	97.2	617695
TBC1D24	172.2	100.0	100.0	615338
TBCD	147.0	96.2	94.4	617193
TBCE	148.9	99.8	97.3	241410
TCF4	132.4	100.0	99.8	610954
TDP2	198.6	100.0	99.4	616949
TOE1	170.4	100.0	100.0	614969
TPP1	146.7	100.0	100.0	204500
TREX1	244.5	100.0	100.0	225750
TRPM3	134.3	100.0	99.5	-
TRPM6	154.4	99.9	99.5	602014

TSC1	136.7	99.8	98.7	607341;191100
TSC2	140.9	100.0	99.6	613254
TSEN15	96.7	99.8	97.5	617026
TSEN2	119.0	100.0	99.6	612389
TSEN54	113.9	96.3	94.3	277470
TUBA1A	77.0	99.9	97.0	611603
TUBB2A	65.8	97.0	95.7	615763
TUBB2B	65.7	100.0	99.5	610031
TUBB4A	76.9	95.9	94.0	612438
TUBG1	178.8	100.0	100.0	615412
UBA5	93.7	97.8	86.8	617132
UBE3A	95.8	99.1	94.8	105830
UBTF	129.1	100.0	99.4	617672
UGP2	154.7	99.0	98.6	618744
VPS53	133.5	91.5	90.7	615851
WDR26	119.5	99.0	96.6	617616
WDR45	77.0	96.4	89.7	300894
WWOX	138.2	100.0	100.0	616211
XK	83.0	99.8	98.1	300842
YWHAG	212.8	100.0	100.0	617665
ZEB2	158.7	99.9	99.1	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.

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