

WES EPILEPSY DG 2.16

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AARS	103.7	100.0	99.5	613287;616339
ABAT	83.2	99.9	98.3	613163
ABCC8	125.8	100.0	99.9	240800
ACTB	80.5	100.0	99.7	243310
ACY1	118.5	100.0	98.6	609924
ADSL	138.6	99.2	98.6	103050
ALDH7A1	66.7	93.5	86.1	266100
ALG1	46.5	53.2	50.2	608540
ALG11	129.3	96.8	96.3	613661
ALG13	77.3	98.5	92.1	300884
ALG3	106.5	100.0	99.9	601110
ALG6	101.6	99.1	95.6	603147
AMACR	157.7	100.0	100.0	614307
AMPD2	132.3	100.0	99.9	615809
AMT	142.7	100.0	100.0	605899
ANKRD11	119.6	99.2	97.1	148050
AP3B2	125.6	99.4	97.6	617276
APOPT1	78.3	82.0	81.5	220110
ARHGEF9	51.5	76.1	71.3	300607
ARID1B	139.6	99.5	99.2	135900
ARX	49.3	87.3	79.2	308350
ASAH1	125.7	99.3	97.2	159950
ASL	123.6	100.0	98.5	207900
ASNS	82.8	98.6	92.2	615574
ASXL3	138.1	99.7	99.1	615485

ATP1A2	161.7	100.0	99.5	104290
ATP1A3	159.8	100.0	100.0	614820
ATP6AP2	44.9	88.4	64.2	300423
ATP7A	111.2	99.5	96.7	309400
ATRX	89.2	99.1	95.5	301042
AUTS2	130.6	99.5	97.7	615834
BOLA3	48.1	99.9	92.5	614299
BRAT1	142.0	100.0	99.3	614498
BTD	126.6	99.9	99.7	253260
CACNA1A	92.4	97.8	94.7	141500
CACNA1E	120.9	99.8	99.2	618285
CACNA2D2	126.0	95.8	94.0	-
CACNB4	97.8	97.2	95.5	607682
CAD	136.7	99.9	99.2	616457
CASK	85.1	99.5	94.5	300749
CDKL5	100.0	95.1	93.1	300672
CHD2	123.9	99.3	99.0	615369
CHRNA2	174.4	100.0	100.0	610353
CHRNA4	109.8	99.9	99.2	600513
CHRNB2	160.6	99.7	98.0	605375
CIC	72.7	64.7	63.3	617600
CLCN4	105.7	99.9	98.9	300114
CLDN16	126.6	100.0	100.0	248250
CLDN19	125.4	99.1	95.1	248190
CLN3	114.7	92.6	91.9	204200
CLN5	138.7	99.9	98.8	256731
CLN6	132.3	100.0	99.9	601780
CLN8	144.5	83.5	83.5	610003
CNNM2	199.8	100.0	100.0	613882
CNTN2	123.7	92.7	92.7	615400

CNTNAP2	127.1	100.0	99.8	610042
COL4A1	95.7	99.6	97.3	175780
COL4A3BP	133.1	99.7	97.8	616351
COLGALT1	149.5	97.8	92.1	618360
COQ2	103.5	97.6	97.1	607426
COQ4	105.0	91.3	90.2	616276
COQ8A	161.8	100.0	99.9	612016
CPA6	109.1	99.4	96.9	614417
CPS1	133.8	100.0	99.9	237300
CPT2	139.2	98.3	98.2	600649
CSTB	70.0	99.3	90.9	254800
CTSD	171.0	99.8	97.8	610127
CTSF	107.0	91.3	81.8	615362
CUL4B	78.0	97.6	89.3	300354
CUX2	120.9	99.9	99.3	618141
D2HGDH	142.0	100.0	99.4	600721
DCX	90.5	99.9	98.4	300067
DDX3X	73.5	86.2	82.9	300958
DENND5A	99.2	99.8	98.9	617281
DEPDC5	124.5	99.9	99.7	604364
DHDDS	81.0	97.1	93.8	617836
DLAT	100.2	99.8	99.2	245348
DNAJC5	188.2	100.0	100.0	162350
DNM1	139.8	94.7	92.3	616346
DOCK7	120.7	99.6	97.8	615730
DPAGT1	87.5	100.0	99.9	608093
DPM1	134.7	95.2	88.2	608799
DPM2	88.5	99.8	97.6	615042
DPYD	141.6	99.5	96.4	274270
DPYS	117.5	100.0	99.8	222748

DYNC1H1	140.6	100.0	99.6	614563
DYRK1A	130.4	100.0	99.9	614104
EEF1A2	188.3	100.0	100.0	616409
EFHC1	117.2	93.0	90.7	607631;254770
EGF	110.5	100.0	99.7	611718
EHMT1	127.7	94.6	94.2	610253
EPM2A	116.5	90.9	88.8	254780
EXOSC3	125.1	96.4	87.8	614678
FA2H	92.7	98.8	92.5	612319
FARS2	161.9	100.0	100.0	614946
FGD1	86.7	98.4	93.0	305400
FGF12	100.4	100.0	99.9	617166
FLNA	142.7	100.0	99.9	300049
FOLR1	107.4	100.0	99.9	613068
FOXP1	142.0	97.8	88.7	613454
FOXRED1	121.0	99.8	98.2	252010
FRMPD4	108.4	99.6	97.2	300983
FRRS1L	99.1	85.5	79.1	616981
FXRD2	108.1	100.0	100.0	154020
GABRA1	164.2	100.0	99.8	615744
GABRB3	131.7	99.5	97.8	617113
GABRG2	126.4	91.1	89.7	611277
GAMT	112.5	98.3	91.5	612736
GCK	138.6	100.0	100.0	602485
GCSH	32.1	88.4	69.8	605899
GLDC	59.2	90.6	79.2	605899
GLRA1	96.8	100.0	99.7	149400
GLRB	104.6	99.5	94.5	614619
GLUD1	65.5	98.1	87.5	606762
GNAO1	152.8	93.8	93.8	615473

GOSR2	102.6	95.8	93.7	614018
GPC3	75.7	98.7	92.7	312870
GPHN	144.7	99.9	98.8	615501
GRIA3	82.4	98.6	92.1	300699
GRIN1	166.1	100.0	99.9	614254
GRIN2A	131.2	100.0	100.0	245570
GRIN2B	158.0	99.8	99.0	613970;616139
GRIN2D	82.8	91.9	79.5	617162
GRN	174.1	100.0	100.0	614706
HACE1	136.2	99.9	99.1	616756
HADH	111.1	99.3	98.8	609975
HCFC1	104.4	99.3	95.8	309541
HCN1	137.5	100.0	99.7	615871
HLCS	142.3	100.0	100.0	253270
HNRNPU	149.9	99.9	99.3	617391
HSD17B10	92.4	100.0	98.4	300438
HSD17B4	109.4	96.3	93.6	261515
ICK	110.0	99.8	99.3	617924
IDH2	98.5	100.0	99.6	613657
IER3IP1	106.3	94.3	82.8	614231
IFIH1	110.8	99.8	98.2	615846
IQSEC2	73.1	95.8	87.9	309530
ITPA	130.2	100.0	100.0	616647
JAM3	126.9	100.0	99.9	613730
KANSL1	139.1	99.8	98.6	610443
KATNB1	154.2	100.0	100.0	616212
KCNA1	150.1	100.0	100.0	160120
KCNA2	126.3	100.0	99.7	616366
KCNB1	129.7	100.0	99.7	616056
KCNC1	170.9	100.0	100.0	616187

KCNH1	148.4	98.7	98.3	611816
KCNJ10	148.6	89.2	88.1	612780
KCNJ11	199.7	100.0	100.0	601820
KCNMA1	102.3	94.8	93.4	609446
KCNQ2	118.3	91.5	90.2	613720
KCNQ3	110.4	99.9	98.7	121201
KCNT1	131.2	96.0	95.1	614959
KCTD7	154.9	95.0	95.0	611726
KDM5C	102.8	99.5	97.0	300534
KPTN	145.7	100.0	100.0	615637
LAMB1	142.9	100.0	99.7	615191
LGI1	133.3	98.3	97.2	600512
LIAS	125.3	99.9	98.7	614462
MBD5	147.7	99.9	99.8	156200
MECP2	124.8	100.0	98.5	312750
MED12	85.1	99.5	95.5	309520
MEF2C	127.9	99.4	95.5	613443
MFSD8	121.3	100.0	99.6	610951
MLC1	96.7	100.0	99.9	604004
MOCS1	91.2	98.8	95.7	252150
MOCS2	137.7	99.6	99.5	252160
MPDU1	102.4	100.0	99.6	609180
MPDZ	128.3	99.6	98.2	615219
MTHFR	114.9	98.2	96.4	236250
MTOR	112.0	99.9	99.1	616638
MTRR	131.1	100.0	99.0	236270
NACC1	169.5	100.0	100.0	617393
NANS	97.2	99.9	98.4	610442
NDUFA1	184.9	99.9	99.2	252010
NDUFA11	116.0	99.8	97.4	252010

NDUF1AF1	98.5	100.0	99.9	252010
NDUF1AF2	54.1	94.3	82.0	252010
NDUF1AF3	141.0	100.0	99.9	252010
NDUF1AF4	98.3	99.2	94.5	252010
NDUF1AF5	124.9	99.9	99.1	252010
NDUFB3	23.3	89.7	62.5	252010
NDUFB9	105.2	97.8	93.3	252010
NDUFS1	143.5	99.9	99.8	252010
NDUFS2	100.1	100.0	100.0	252010
NDUFS3	124.8	90.7	90.5	252010
NDUFS4	144.5	100.0	99.7	252010
NDUFS6	111.9	100.0	100.0	252010
NDUFV1	141.7	99.9	98.8	252010
NDUFV2	74.2	92.4	77.3	252010
NECAP1	102.3	100.0	100.0	615833
NEDD4L	93.7	72.3	71.5	617201
NEU1	141.3	99.3	96.4	256550
NEXMIF	132.0	100.0	99.5	300912
NGLY1	135.4	100.0	99.7	615273
NHLRC1	169.7	100.0	100.0	254780
NPRL2	138.7	100.0	100.0	617116
NPRL3	120.3	100.0	99.8	617118
NRXN1	141.6	97.6	97.3	614325
NUBPL	102.0	98.9	95.5	252010
OCLN	173.9	100.0	100.0	251290
OFD1	51.9	85.8	70.8	311200
OPHN1	78.3	98.9	95.0	300486
PAFAH1B1	77.0	92.0	82.8	607432
PAK3	85.3	98.6	93.7	300558
PC	155.4	99.9	98.7	266150

PCDH19	176.6	99.9	98.9	300088
PDHA1	85.3	98.9	95.4	312170
PDHB	111.4	99.2	97.2	614111
PDP1	129.1	100.0	100.0	608782
PDX1	72.9	99.1	95.2	245349
PET100	87.9	98.0	87.6	220110
PEX1	127.9	99.9	99.3	214100
PEX10	113.3	99.9	97.4	614870
PEX12	120.6	100.0	100.0	614859
PEX13	179.6	100.0	100.0	614883
PEX14	130.5	99.8	97.8	614887
PEX16	140.8	98.6	94.8	614876
PEX19	84.9	100.0	98.9	614886
PEX26	94.3	100.0	99.6	614872
PEX3	113.9	99.9	99.2	614882
PEX5	107.9	100.0	99.2	600414
PEX6	106.5	98.5	92.0	614862
PGAP3	70.3	63.5	59.9	615716
PHF6	60.3	98.2	87.9	301900
PHGDH	106.6	100.0	99.3	601815
PIGA	70.9	92.9	84.0	300868
PIGN	106.3	93.6	91.1	614080
PIGO	144.5	100.0	99.9	614749
PIGP	89.3	94.8	86.0	617599
PIGT	159.3	98.1	98.1	615398
PLA2G6	111.9	99.8	98.2	256600
PLCB1	134.9	100.0	99.7	613722
PLP1	112.8	99.7	97.7	312080
PLPBP	95.3	99.6	95.3	617290
PMM2	127.7	100.0	99.7	212065

PNKP	109.0	100.0	99.9	613402
PNPO	74.4	100.0	99.3	610090
POLG	113.9	100.0	99.6	203700
PPP2R1A	129.1	91.6	91.6	616362
PPP2R5D	136.6	100.0	100.0	616355
PPT1	136.6	90.2	89.2	256730
PQBP1	163.5	100.0	100.0	309500
PRF1	138.1	91.2	90.6	603553
PRICKLE1	100.0	100.0	99.8	612437
PRRT2	111.8	100.0	99.0	605751
PSAP	98.1	100.0	99.3	611722
PUM1	126.9	100.0	99.5	617931
PURA	207.1	99.5	96.9	616158
PYCR2	116.5	99.7	96.9	616420
QARS	129.2	100.0	99.8	615760
RAB39B	102.0	100.0	99.9	300271
RARS2	104.0	100.0	99.4	611523
RNASEH2A	129.8	100.0	99.7	610333
RNASEH2B	100.8	98.9	95.2	610181
RNASEH2C	281.7	100.0	100.0	610329
ROGDI	127.6	100.0	99.4	226750
RPS6KA3	87.8	98.3	93.0	303600
RRM2B	143.9	99.9	99.4	612075
SAMHD1	133.4	99.8	98.5	612952
SCARB2	105.8	99.8	99.1	254900
SCN1A	121.4	100.0	99.1	607208
SCN1B	169.7	99.9	98.1	604233
SCN2A	132.4	99.6	97.7	613721
SCN3A	138.7	99.9	99.1	617938;617935
SCN8A	154.3	100.0	99.7	614558

SEPSECS	159.6	100.0	99.6	613811
SERPINI1	101.2	99.8	97.4	604218
SHANK3	123.9	97.5	91.6	606232
SIK1	118.5	99.6	96.7	616341
SLC12A5	111.9	86.1	84.1	616645
SLC13A5	141.9	100.0	99.9	608305
SLC16A1	138.1	100.0	99.2	610021
SLC19A3	134.6	100.0	99.9	607483
SLC1A2	97.1	99.3	97.2	617105
SLC25A1	103.2	99.3	95.1	615182
SLC25A15	146.8	97.9	93.6	238970
SLC25A22	123.8	100.0	99.1	609304
SLC2A1	148.9	92.8	92.8	606777
SLC35A2	104.8	99.8	98.1	300896
SLC6A1	126.0	100.0	100.0	616421
SLC6A8	53.5	96.1	83.8	300352
SLC9A6	101.0	98.6	94.3	300243
SMARCA2	105.9	96.8	95.9	601358
SMC1A	87.8	99.9	97.8	300590
SMS	63.0	88.9	73.1	309583
SNAP25	119.0	99.9	99.7	-
SPATA5	139.5	100.0	99.8	616577
SPTAN1	112.0	99.1	98.3	613477
ST3GAL3	134.7	100.0	99.5	615006
ST3GAL5	101.8	89.0	84.9	609056
STX1B	157.7	100.0	100.0	616172
STXBP1	103.7	96.8	96.4	612164
SUOX	167.2	100.0	100.0	272300
SYN1	66.6	90.6	79.1	300491
SYNGAP1	140.7	98.4	97.7	612621

SYNJ1	126.6	99.9	98.5	617389
SYP	79.9	99.9	98.1	300802
SZT2	135.6	99.6	99.4	615476
TANGO2	127.3	100.0	100.0	616878
TBC1D23	92.7	99.2	95.4	617695
TBC1D24	177.7	100.0	100.0	615338
TBCD	136.2	98.2	94.3	617193
TBCE	116.4	98.7	94.7	241410
TCF4	109.2	100.0	99.8	610954
TDP2	173.0	99.9	99.4	616949
TOE1	141.1	100.0	99.8	614969
TPP1	123.7	100.0	99.9	204500
TREX1	233.4	100.0	100.0	225750
TRPM6	126.6	99.9	99.1	602014
TSC1	112.5	99.6	98.2	607341;191100
TSC2	140.5	100.0	99.9	613254
TSEN15	89.8	99.7	96.4	617026
TSEN2	95.6	99.9	98.9	612389
TSEN54	114.4	99.4	96.8	277470
TUBA1A	77.6	99.8	97.1	611603
TUBB2A	77.1	99.7	97.2	615763
TUBB2B	78.2	100.0	99.7	610031
TUBB4A	101.2	97.1	95.6	612438
TUBG1	154.3	100.0	100.0	615412
UBA5	79.9	97.7	86.6	617132
UBE3A	81.5	98.6	93.3	105830
VPS53	111.3	91.1	89.6	615851
WDR26	98.2	99.5	97.4	617616
WDR45	68.7	96.8	88.9	300894
WVOX	116.1	100.0	99.9	616211

XK	85.4	100.0	99.4	300842
YWHAG	167.1	100.0	100.0	617665
ZEB2	140.1	99.7	98.4	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