

WES EPILEPSY DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AARS	124.3	100.0	99.6	613287;616339
ABAT	92.7	100.0	99.5	613163
ABCC8	146.6	100.0	99.9	240800
ACTB	129.0	99.1	94.2	243310
ACY1	132.8	99.9	98.3	609924
ADSL	183.6	99.2	99.1	103050
ALDH7A1	77.1	93.7	85.4	266100
ALG1	50.9	53.6	48.8	608540
ALG11	139.6	96.7	96.0	613661
ALG13	86.7	98.7	94.1	300884
ALG3	132.9	100.0	100.0	601110
ALG6	96.4	96.0	93.3	603147
AMACR	157.9	100.0	100.0	614307
AMPD2	135.5	99.9	99.2	615809
AMT	173.1	100.0	100.0	605899
ANKRD11	96.3	97.4	94.1	148050
AP3B2	135.1	97.5	94.2	617276
APOPT1	63.8	81.4	78.1	220110
ARHGEF9	60.2	76.4	74.4	300607
ARID1B	156.7	94.3	89.5	135900
ARX	29.1	75.8	59.5	308350
ASAH1	105.9	97.6	92.1	159950
ASL	114.4	99.9	98.8	207900
ASXL3	162.9	99.8	98.9	615485
ATP1A2	190.8	100.0	99.6	104290

ATP1A3	177.3	100.0	100.0	614820
ATP6AP2	46.1	81.2	55.6	300423
ATP7A	133.2	99.7	97.8	309400
ATRX	82.6	98.2	92.2	301042
AUTS2	110.3	96.9	95.5	615834
BOLA3	50.1	92.3	81.7	614299
BRAT1	108.5	99.8	97.4	614498
BTD	166.6	100.0	99.9	253260
CACNA1A	87.8	92.7	89.1	141500
CACNA1E	139.3	99.8	99.3	-
CACNA2D2	135.2	93.7	92.6	-
CACNB4	106.1	96.3	94.6	607682
CAD	158.9	100.0	99.7	616457
CASK	92.3	98.7	93.7	300749
CDKL5	114.4	94.9	91.8	300672
CHD2	137.7	99.3	98.5	615369
CHRNA2	229.3	100.0	100.0	610353
CHRNA4	142.1	96.7	95.8	600513
CHRNB2	247.5	98.4	94.6	605375
CLCN4	123.0	100.0	99.8	300114
CLDN16	136.3	100.0	99.9	248250
CLDN19	123.7	98.2	93.7	248190
CLN3	114.9	92.5	90.7	204200
CLN5	146.1	98.2	92.2	256731
CLN6	131.6	98.9	95.3	601780
CLN8	163.9	83.5	83.5	610003
CNNM2	188.4	100.0	99.2	613882
CNTN2	125.4	92.7	92.6	615400
CNTNAP2	148.0	100.0	99.9	610042
COL4A1	92.8	97.9	94.0	175780

COL4A3BP	121.6	98.3	92.7	616351
COQ2	89.3	96.1	93.2	607426
COQ4	89.8	88.4	84.9	616276
COQ8A	134.3	100.0	99.1	612016
CPA6	118.3	99.8	98.5	614417
CPS1	143.8	100.0	99.8	237300
CPT2	162.8	97.2	95.4	600649
CSTB	82.5	97.1	82.7	254800
CTSD	163.7	98.0	95.3	610127
CTSF	112.8	84.2	80.2	615362
CUL4B	72.8	98.0	88.5	300354
CUX2	85.6	99.8	97.9	-
D2HGDH	134.5	97.5	95.2	600721
DCX	113.2	100.0	99.7	300067
DDX3X	80.5	85.9	82.1	300958
DENND5A	123.0	99.8	97.9	617281
DEPDC5	148.3	99.8	99.3	604364
DHDDS	93.5	97.8	94.8	617836
DLAT	91.6	99.1	96.0	245348
DNAJC5	200.2	100.0	99.9	162350
DNM1	156.7	89.3	87.5	616346
DOCK7	114.4	97.9	95.6	615730
DPAGT1	110.7	100.0	100.0	608093
DPM1	131.1	91.7	86.7	608799
DPM2	102.1	100.0	99.4	615042
DPYD	158.3	95.6	93.7	274270
DPYS	133.5	100.0	99.5	222748
DYNC1H1	179.8	100.0	99.6	614563
DYRK1A	159.6	100.0	100.0	614104
EEF1A2	177.7	98.8	93.8	616409

EFHC1	136.3	92.2	91.5	607631;254770
EGF	135.2	100.0	99.8	611718
EHMT1	141.4	94.0	92.9	610253
EPM2A	110.1	86.2	84.0	254780
EXOSC3	88.5	97.3	89.4	614678
FA2H	94.1	87.9	79.9	612319
FARS2	207.7	100.0	100.0	614946
FGD1	85.7	92.7	86.5	305400
FGF12	95.5	99.6	96.3	617166
FLNA	138.1	100.0	99.5	300049
FOLR1	150.4	100.0	100.0	613068
FOXG1	157.5	84.8	81.0	613454
FOXRED1	136.6	100.0	99.6	252010
FRMPD4	114.1	99.7	98.0	300983
FRRS1L	103.1	68.3	63.4	616981
FXVD2	96.7	99.8	99.2	154020
GABRA1	179.5	100.0	100.0	615744
GABRB3	140.5	98.1	93.7	617113
GABRG2	137.9	91.1	90.3	611277
GAMT	93.5	90.9	80.7	612736
GCK	141.4	100.0	100.0	602485
GCSH	34.2	83.1	67.8	605899
GLDC	78.9	90.6	82.7	605899
GLRA1	123.3	100.0	100.0	149400
GLRB	98.6	96.6	88.8	614619
GLUD1	74.5	94.4	84.3	606762
GNAO1	167.8	93.8	93.8	615473
GOSR2	127.2	95.9	95.0	614018
GPC3	85.1	98.6	92.6	312870
GPHN	167.2	98.4	96.9	615501

GRIA3	98.2	99.3	94.7	300699
GRIN1	150.7	100.0	99.5	614254
GRIN2A	159.1	100.0	100.0	245570
GRIN2B	189.4	99.9	99.3	613970;616139
GRIN2D	73.1	69.1	62.1	617162
GRN	184.5	100.0	100.0	614706
HADH	110.8	98.0	95.1	609975
HCFC1	105.8	99.3	96.1	309541
HCN1	122.4	99.9	97.8	615871
HLCS	172.8	100.0	100.0	253270
HNRNPU	126.6	99.3	97.9	617391
HSD17B10	117.1	100.0	99.2	300438
HSD17B4	95.1	93.9	90.8	261515
ICK	131.6	99.9	98.2	-
IDH2	103.5	99.6	96.9	613657
IER3IP1	73.0	93.2	82.2	614231
IFIH1	113.5	99.6	97.1	615846
IQSEC2	61.2	92.1	82.5	309530
ITPA	120.2	100.0	100.0	616647
JAM3	158.6	100.0	100.0	613730
KANSL1	172.3	99.9	99.2	610443
KATNB1	141.7	100.0	100.0	616212
KCNA1	167.0	100.0	99.7	160120
KCNA2	157.7	100.0	99.9	616366
KCNB1	145.0	100.0	99.9	616056
KCNC1	199.2	100.0	100.0	616187
KCNH1	185.8	98.7	98.7	611816
KCNJ10	213.4	89.3	89.1	612780
KCNJ11	299.5	100.0	100.0	601820
KCNMA1	120.4	94.4	93.2	609446

KCNQ2	85.4	90.1	86.5	613720
KCNQ3	110.7	98.9	95.5	121201
KCNT1	112.0	95.3	92.3	614959
KCTD7	166.7	95.0	95.0	611726
KDM5C	112.6	97.9	95.1	300534
KPTN	112.1	100.0	99.9	615637
LAMB1	169.8	100.0	99.6	615191
LGI1	163.8	97.8	94.8	600512
LIAS	133.7	99.5	97.1	614462
MBD5	196.2	99.9	99.6	156200
MECP2	87.3	99.1	93.1	312750
MED12	105.7	98.0	94.8	309520
MEF2C	137.7	97.9	93.5	613443
MFSD8	125.1	99.9	98.4	610951
MLC1	103.4	100.0	99.8	604004
MOCS1	87.4	98.4	93.0	252150
MOCS2	139.5	99.6	99.6	252160
MPDU1	111.8	100.0	99.7	609180
MPDZ	149.0	98.7	96.6	615219
MTHFR	126.1	98.4	97.2	236250
MTOR	140.0	100.0	99.8	616638
MTRR	139.1	100.0	99.2	236270
NACC1	167.7	100.0	99.9	617393
NANS	106.1	100.0	99.9	610442
NDUFA1	166.8	100.0	99.6	252010
NDUFA11	86.9	99.5	95.8	252010
NDUFAF1	115.6	100.0	100.0	252010
NDUFAF2	58.6	85.7	70.9	252010
NDUFAF3	120.8	100.0	100.0	252010
NDUFAF4	79.4	98.9	91.8	252010

NDUFAF5	95.7	98.8	94.5	252010
NDUFB3	22.6	91.9	59.2	252010
NDUFB9	120.1	99.8	97.4	252010
NDUFS1	132.2	99.8	98.6	252010
NDUFS2	117.8	100.0	100.0	252010
NDUFS3	142.4	90.7	90.6	252010
NDUFS4	147.3	100.0	99.1	252010
NDUFS6	119.1	99.9	99.4	252010
NDUFV1	136.7	99.7	97.8	252010
NDUFV2	69.5	78.7	53.9	252010
NECAP1	116.9	100.0	99.9	615833
NEDD4L	105.2	71.7	70.5	617201
NEXMIF	139.2	99.9	99.0	300912
NGLY1	128.0	100.0	99.5	615273
NHLRC1	174.2	100.0	100.0	254780
NPRL2	168.5	100.0	100.0	617116
NPRL3	127.5	100.0	99.7	617118
NRXN1	160.9	96.8	95.7	614325
NUBPL	89.8	92.9	85.9	252010
OCLN	220.7	100.0	100.0	251290
OFD1	51.5	84.0	67.8	311200
OPHN1	89.0	99.1	96.2	300486
PAFAH1B1	105.2	89.1	81.4	607432
PAK3	82.8	97.6	91.8	300558
PC	149.3	97.7	94.6	266150
PCDH19	224.1	100.0	99.3	300088
PDHA1	109.8	98.1	92.1	312170
PDHB	133.0	99.3	96.8	614111
PDP1	209.6	100.0	100.0	608782
PDX1	35.4	89.0	72.1	245349

PET100	94.5	88.8	74.8	220110
PEX1	115.8	97.7	95.4	214100
PEX10	111.8	96.1	90.1	614870
PEX12	168.3	100.0	100.0	614859
PEX13	197.6	99.8	98.7	614883
PEX14	149.0	99.7	97.5	614887
PEX16	137.0	97.1	93.1	614876
PEX19	92.9	99.9	99.2	614886
PEX26	76.4	100.0	99.8	614872
PEX3	98.1	99.1	94.3	614882
PEX5	111.7	99.9	98.3	600414
PEX6	94.5	90.4	86.1	614862
PGAP3	72.8	62.5	58.0	615716
PHF6	62.8	92.6	83.7	301900
PHGDH	115.6	100.0	99.8	601815
PIGA	90.5	90.4	81.3	300868
PIGN	111.3	92.6	87.1	614080
PIGO	147.0	100.0	99.9	614749
PIGP	101.4	91.6	83.8	617599
PIGT	171.3	98.1	98.0	615398
PLA2G6	117.5	99.9	98.4	256600
PLCB1	142.8	100.0	99.7	613722
PLP1	129.2	100.0	99.4	312080
PLPBP	112.6	99.1	92.6	617290
PMM2	141.1	99.9	99.4	212065
PNKP	93.0	99.8	97.7	613402
PNPO	66.4	100.0	98.3	610090
POLG	114.4	100.0	99.5	203700
PPP2R1A	134.0	91.7	91.6	616362
PPT1	144.5	90.0	87.3	256730

PQBP1	186.1	100.0	100.0	309500
PRF1	122.5	91.2	90.8	603553
PRICKLE1	117.3	100.0	100.0	612437
PRRT2	78.9	99.9	98.4	605751
PSAP	114.4	99.9	99.0	611722
PUM1	158.0	100.0	99.9	617931
PURA	121.8	94.5	87.2	616158
PYCR2	127.6	100.0	97.6	616420
QARS	166.6	100.0	100.0	615760
RAB39B	113.0	100.0	99.7	300271
RARS2	107.2	100.0	99.1	611523
RNASEH2A	142.1	100.0	99.9	610333
RNASEH2B	103.8	93.2	87.5	610181
RNASEH2C	209.2	100.0	99.9	610329
ROGDI	112.2	97.9	95.3	226750
RPS6KA3	79.3	94.2	83.3	303600
RRM2B	128.6	99.7	97.5	612075
SAMHD1	127.9	99.6	96.6	612952
SCARB2	121.2	100.0	99.9	254900
SCN1A	135.2	99.6	98.0	607208
SCN1B	168.3	97.1	96.1	604233
SCN2A	156.7	99.0	96.4	613721
SCN8A	198.3	100.0	99.7	614558
SEPSECS	159.3	100.0	100.0	613811
SIK1	85.6	97.0	92.4	616341
SLC12A5	125.9	85.4	81.6	616645
SLC13A5	164.1	100.0	100.0	608305
SLC16A1	157.1	99.9	98.8	610021
SLC19A3	186.4	100.0	99.9	607483
SLC1A2	128.0	99.9	99.4	617105

SLC25A1	71.0	92.2	87.0	615182
SLC25A15	192.5	98.8	95.0	238970
SLC25A22	108.7	99.5	96.9	609304
SLC2A1	190.1	92.9	92.8	606777
SLC35A2	108.8	99.7	96.8	300896
SLC6A1	143.6	100.0	100.0	616421
SLC6A8	56.5	89.8	79.1	300352
SLC9A6	104.2	97.6	91.3	300243
SMARCA2	113.8	95.7	93.7	601358
SMC1A	99.4	99.9	98.8	300590
SMS	67.8	88.3	73.9	309583
SNAP25	133.7	100.0	99.9	-
SPTAN1	125.5	99.1	98.6	613477
ST3GAL3	144.5	100.0	99.9	615006
ST3GAL5	121.9	84.4	84.2	609056
STX1B	152.2	100.0	98.4	616172
STXBP1	124.5	96.8	96.8	612164
SUOX	212.6	100.0	100.0	272300
SYN1	64.2	74.0	63.2	300491
SYNGAP1	141.3	98.4	98.0	612621
SYNJ1	127.2	99.3	96.1	617389
SYP	72.0	99.8	94.0	300802
SZT2	149.5	99.5	99.2	615476
TANGO2	145.3	100.0	100.0	616878
TBC1D23	86.0	95.7	91.5	617695
TBC1D24	179.2	100.0	100.0	615338
TBCE	128.0	99.9	98.2	241410
TCF4	128.0	99.9	99.5	610954
TDP2	165.1	99.9	98.8	616949
TOE1	165.1	100.0	100.0	614969

TPP1	146.3	100.0	100.0	204500
TREX1	242.4	100.0	100.0	225750
TRPM6	151.1	99.8	98.7	602014
TSC1	128.8	99.8	98.8	607341;191100
TSC2	131.2	100.0	99.0	613254
TSEN15	74.2	99.0	93.6	617026
TSEN2	123.8	100.0	99.8	612389
TSEN54	82.9	95.9	92.9	277470
TUBA1A	113.2	99.9	97.8	611603
TUBB2A	109.9	96.7	95.6	615763
TUBB2B	100.0	100.0	100.0	610031
TUBB4A	121.2	96.0	95.3	612438
TUBG1	164.2	100.0	100.0	615412
UBA5	75.4	94.1	77.1	617132
UBE3A	89.8	97.8	91.4	105830
VPS53	129.2	91.4	90.4	615851
WDR45	75.0	97.4	90.1	300894
WWOX	130.9	100.0	99.7	616211
XK	96.8	99.9	99.1	300842
YWHAG	226.6	100.0	100.0	617665
ZEB2	157.0	99.8	98.8	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