WES NEUROPATHIES DG 3.1

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
AAAS	117.1	100.0	99.9	231550
AARS1	124.9	100.0	99.9	613287
ABCA1	115.8	99.9	99.1	205400
ABCD1	78.1	75.8	71.6	300100
ABHD12	94.6	91.2	85.2	612674
ADPRS	159.0	100.0	99.8	MIM pheno ID 618170
AFG3L2	115.3	95.0	91.1	No OMIM phenotype
AGRN	139.7	96.9	92.6	615120
AGTPBP1	144.2	96.0	94.1	618276
AIFM1	117.3	99.9	98.8	310490
AMACR	159.5	100.0	100.0	614307
APTX	122.7	94.9	92.4	208920
ARHGEF10	132.1	99.8	98.0	608236
ARHGEF28	117.2	99.2	94.4	-
ARSA	126.4	100.0	99.8	250100
ATAD3A	84.8	91.9	83.2	617183
ATL1	180.1	100.0	99.7	613708
ATL3	147.6	99.8	98.3	615632
ATP13A2	138.1	100.0	99.5	No OMIM phenotype
ATP1A1	143.2	100.0	100.0	618036
ATP7A	134.6	99.0	96.9	304150
BAG3	184.0	100.0	100.0	613881;612954
BICD2	137.0	100.0	99.7	615290
BSCL2	113.7	100.0	100.0	600794
C12orf65	122.8	99.8	98.5	613559;615035

C19orf12	122.6	100.0	99.8	615043;614298
C1orf194	66.7	100.0	99.6	-
CADM3	104.1	100.0	99.9	No OMIM phenotype
CCT5	146.2	100.0	99.7	256840
CHCHD10	25.2	59.1	43.9	615911;616209;615048
CNTNAP1	173.7	100.0	99.8	618186
COA3	156.1	100.0	100.0	220110
COA7	142.6	100.0	100.0	618387
COL6A5	163.1	99.9	99.5	28073787
COX20	72.8	97.8	88.3	220110
COX6A1	177.4	100.0	99.5	616039
CTDP1	129.4	88.4	84.3	604168
CYP27A1	180.7	98.9	96.7	213700
DCAF8	110.8	100.0	99.9	610100
DCTN1	126.0	100.0	98.8	607641
DCTN2	102.0	100.0	99.7	-
DGAT2	93.6	99.1	95.5	-
DHTKD1	145.8	99.9	98.9	615025
DNAH10	150.2	99.9	99.4	-
DNAJB2	139.8	100.0	100.0	614881
DNAJB5	140.6	95.7	91.3	-
DNAJC3	148.3	100.0	99.7	616192
DNM1L	139.3	99.9	98.5	PMID: 33387674;31868880;30085106
DNM2	138.9	98.1	94.5	606482
DNMT1	131.2	99.2	99.0	614116
DRP2	92.3	99.1	96.5	-
DST	168.2	95.5	95.0	614653
DYNC1H1	157.5	99.9	99.4	614228
EGR2	152.7	100.0	100.0	145900;607678
ELOVL5	130.0	100.0	99.8	615957

ELP1	149.3	99.8	99.0	223900
EMILIN1	107.7	99.3	89.8	PMID: 31978608;26462740
EXOSC8	91.0	97.9	91.2	616081
EXOSC9	173.4	99.7	97.2	618065
FAM126A	146.4	100.0	99.4	610532
FBLN5	121.2	91.8	91.8	614434;608895;219100
FBXO38	188.8	99.9	99.3	615575
FGD4	120.6	99.9	99.4	609311
FIG4	190.3	100.0	99.8	611228
FLVCR1	153.2	100.0	98.9	609033
FXN	72.3	95.5	80.1	MIM pheno ID: 229300
GALC	117.3	99.8	98.3	245200
GAN	177.4	100.0	99.6	256850
GARS1	154.4	99.9	99.1	601472;600794
GBE1	200.1	100.0	99.6	263570;232500
GBF1	142.1	98.3	98.0	PMID: 32937143
GDAP1	185.1	99.8	99.3	608340;214400;607831;607706
GJB1	169.1	100.0	100.0	302800
GJB3	250.6	100.0	100.0	133200;220290;612644
GLA	81.9	91.1	88.2	301500
GLE1	103.6	100.0	100.0	611890
GNB4	194.6	100.0	100.0	615185
GNE	132.7	100.0	99.7	PMID: 31167812
GSN	117.2	95.8	93.5	105120
HADHA	87.5	97.2	91.6	609015
HADHB	90.7	98.8	89.7	609015
HARS1	152.0	100.0	100.0	614504;616625
HINT1	62.4	98.3	89.3	137200
HK1	140.2	100.0	100.0	605285
HMBS	103.3	99.9	99.4	176000

HOXD10	166.0	100.0	100.0	192950
HSD17B4	130.0	95.4	93.1	261515;233400
HSPB1	55.8	98.8	91.6	606595;608634
HSPB3	275.4	100.0	100.0	613376
HSPB8	187.6	100.0	100.0	158590;608673
IFRD1	155.4	99.7	98.6	-
IGHMBP2	118.2	98.8	95.1	604320
INF2	89.3	86.7	83.8	614455
ITPR3	152.6	100.0	99.7	PMID: 27549087;PMID: 24627108;PMID: 32949214
JAG1	147.4	97.7	96.8	PMID: 32065591
KARS1	133.2	100.0	99.9	613641
KBTBD13	102.9	99.8	95.8	609273
KIF1A	114.7	97.4	95.2	614213
KIF1B	167.4	100.0	99.6	118210;256700;171300
KIF5A	141.4	100.0	99.9	604187
KLC2	129.2	99.2	97.9	No OMIM phenotype
LAMA2	155.8	100.0	99.6	PMID: 25648254;PMID: 24957499
LITAF	130.2	98.2	92.7	601098
LMNA	97.4	97.4	91.9	605588
LRIG3	187.6	99.8	98.8	-
LRSAM1	139.4	100.0	99.9	614436
MAG	161.0	100.0	100.0	No OMIM phenotype
MARS1	114.4	99.7	97.4	616280;615486
MCM3AP	149.9	99.9	99.1	618124
MED25	151.9	100.0	99.8	605589
MFN2	138.2	100.0	99.9	601152;609260
MME	138.2	99.8	98.7	617017;617018
MORC2	141.7	100.0	99.8	616688
MPV17	97.4	100.0	97.2	618400;256800
MPZ	104.4	87.9	84.1	145900;607791;118200;607677;607736

MTMR2	120.1	100.0	99.0	601382
MYH14	114.1	98.4	94.0	614369;600652
MYO1A	126.2	100.0	99.8	-
NAGLU	118.0	92.9	89.9	616491
NARS1	181.6	100.0	100.0	PMID: 32738225
NDRG1	133.0	100.0	100.0	601455
NDUFA9	115.5	99.9	96.5	618247
NEFH	108.0	93.4	84.5	162230
NEFL	153.5	99.9	98.2	607734;607684
NEMF	135.8	100.0	99.2	No OMIM phenotype
NFASC	139.4	100.0	99.9	618356
NGF	224.8	100.0	100.0	608654
NIPA1	191.5	100.0	100.0	600363
NMNAT2	110.5	99.9	98.9	-
NTRK1	138.9	99.8	98.2	256800
PDK3	122.5	99.5	97.2	300905
PDXK	122.8	79.3	76.6	618511
PDYN	138.2	100.0	100.0	610245
PEX1	155.5	99.9	99.4	601539;214100
PEX10	101.8	96.8	89.7	614870;614871
PEX16	158.6	97.9	94.2	614876;614877
PEX7	135.9	87.8	80.7	614879
PHYH	89.1	100.0	99.6	266500
PIEZO2	121.6	100.0	99.5	617146
PLA2G6	112.9	92.2	90.7	256600
PLD3	189.9	99.9	99.1	617770
PLEKHG5	97.3	95.3	91.1	615376;611067
PMM2	145.4	100.0	100.0	212065
PMP2	149.0	100.0	100.0	618279
PMP22	123.5	100.0	100.0	145900;118300;118220

PNKP	112.0	100.0	100.0	613402;616267
PNPT1	63.9	97.7	89.7	614932
POLG	111.5	100.0	99.3	258450;607459;157640
POLG2	235.4	99.6	98.0	PMID: 28078310;PMID: 29625556
POLR3B	157.1	99.9	98.6	PMID: 33417887
PRDM12	123.7	90.8	88.0	616488
PRNP	133.8	100.0	100.0	PMID: 24224623;PMID: 29984897
PRPS1	118.4	86.4	86.4	301835;311070
PRX	142.8	96.0	95.5	145900;614895
PSAP	112.4	100.0	100.0	611722
RAB7A	151.4	100.0	99.9	600882
REEP1	82.5	78.7	76.1	610250;614751
RETREG1	141.2	98.8	95.1	613115
RNF170	146.5	99.6	97.6	608984
SACS	174.6	100.0	99.9	270550
SAMD9L	192.5	100.0	100.0	159550
SBF1	130.0	99.0	97.7	615284
SBF2	130.5	99.9	99.4	604563
SCARB2	121.3	100.0	99.8	PMID: 31407473;PMID: 21670406
SCN10A	148.7	100.0	99.6	615551
SCN11A	144.6	99.8	98.3	615548
SCN9A	163.1	99.3	97.9	133020;243000
SCO2	107.3	100.0	100.0	604377
SCP2	130.8	100.0	99.2	No OMIM phenotype
SCYL1	155.2	100.0	100.0	616719
SEPTIN9	154.2	100.0	99.9	162100
SETX	182.3	100.0	99.8	602433;606002
SGPL1	159.8	100.0	100.0	-
SH3TC2	107.9	100.0	99.7	601596;613353
SIGMAR1	134.5	100.0	100.0	614373;605726

SLC12A6	146.8	100.0	100.0	218000
SLC25A19	94.0	100.0	98.5	MIM pheno ID: 613710;PMID: 31295743;PMID: 19798730
SLC25A46	199.3	99.7	97.3	616505
SLC52A2	170.8	100.0	100.0	614707
SLC52A3	125.5	100.0	100.0	211530
SLC5A7	118.9	100.0	99.9	158580
SORD	100.3	90.3	89.1	618912
SOX10	70.3	99.9	97.9	609136
SPG11	135.0	100.0	99.3	616668
SPTAN1	126.6	99.1	98.6	PMID: 31332438
SPTBN4	91.4	97.3	91.0	617519
SPTLC1	132.2	99.2	95.4	162400
SPTLC2	158.5	100.0	100.0	613640
SPTLC3	156.9	100.0	99.9	-
SURF1	94.9	89.4	88.2	256000;616684
SYT2	98.5	99.9	99.0	616040
TBCE	146.6	99.8	97.5	617207
TDP1	123.5	99.9	99.5	607250
TDRKH	107.2	94.7	94.7	PMID: 30503856
TFG	139.5	96.9	96.3	604484
TRIM2	164.9	93.9	93.3	615490
TRPV4	158.6	100.0	99.9	606071
TTR	151.6	94.6	94.6	105210
TUBB2A	65.8	97.0	95.7	PMID: 32203252;29547997
TUBB3	113.8	98.3	96.9	600638
TWNK	202.8	100.0	100.0	616138
UBA5	93.7	97.8	86.8	No OMIM phenotype
UCHL1	117.1	99.8	92.5	615491
UQCRC1	132.5	99.8	98.4	PMID: 33141179
VCP	122.7	100.0	99.2	616687

VRK1	156.9	99.7	98.5	607596
VWA1	67.0	84.1	76.3	No OMIM phenotype
WARS1	121.4	99.8	98.3	617721
WNK1	156.2	99.9	99.6	201300
XRCC1	130.5	100.0	98.8	No OMIM phenotype
YARS1	131.1	100.0	99.9	608323
ZFYVE26	116.7	100.0	99.1	No OMIM phenotype

Gene symbols used follow HGCN 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 10x 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