

WES NEUROPATHIES (HMSN)* 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
ABHD12	107.0	97.3	88.0	612674
AIFM1	106.2	100.0	99.7	310490
ARHGEF10	132.8	99.8	98.0	608236
ATL1	161.0	99.7	97.9	613708
ATL3	125.0	98.1	93.8	615632
ATP7A	133.2	99.7	97.8	304150
BSCL2	113.5	100.0	100.0	600794
CCT5	164.5	99.9	99.1	256840
COX6A1	180.6	100.0	99.4	616039
CTDP1	105.0	86.6	83.6	604168
DCAF8	124.3	100.0	99.9	610100
DCTN1	131.6	99.7	98.3	607641
DHH	117.7	100.0	100.0	607080
DHTKD1	141.0	99.6	98.2	615025
DNAJB2	102.0	100.0	100.0	614881
DNM2	127.4	97.5	94.4	606482
DNMT1	113.4	99.2	98.3	614116
DYNC1H1	179.8	100.0	99.6	614228
EGR2	124.4	100.0	100.0	145900;607678
ELP1	142.6	99.7	98.1	223900
EXOSC8	80.1	91.8	76.9	616081
FBLN5	119.6	91.8	91.1	608895;219100;614434
FGD4	111.9	99.3	97.3	609311
FIG4	154.9	99.8	98.4	611228

FLVCR1	139.5	99.2	95.8	609033
GAN	190.0	100.0	99.9	256850
GARS	125.7	99.9	98.5	601472;600794
GDAP1	163.1	99.3	96.1	214400;607706;608340;607831
GJB1	229.8	100.0	99.8	302800
GJB3	308.9	100.0	100.0	612644;220290;133200
GNB4	152.6	100.0	99.7	615185
HARS	159.4	100.0	100.0	614504;616625
HINT1	60.0	98.5	88.0	137200
HOXD10	137.7	100.0	99.8	192950
HSPB1	39.7	93.7	81.8	606595;608634
HSPB3	290.9	100.0	100.0	613376
HSPB8	156.4	100.0	100.0	608673;158590
IGHMBP2	107.8	99.3	96.0	604320
INF2	79.2	84.1	81.1	614455
KARS	122.6	100.0	99.3	613641
KIF1A	114.0	99.2	96.1	614213
KIF1B	154.8	100.0	99.5	256700;118210;171300
KIF5A	136.1	100.0	99.9	604187
LITAF	126.3	94.8	91.5	601098
LMNA	89.2	97.9	91.3	605588
LRSAM1	130.4	100.0	99.7	614436
MARS	125.2	99.7	97.3	615486;616280
MED25	103.9	99.1	95.7	605589
MFN2	150.6	100.0	99.9	601152;609260
MME	101.1	98.2	93.6	617017
MPZ	123.4	100.0	99.3	118200;607791;145900;607677;607736
MTMR2	106.6	100.0	99.2	601382
MYH14	102.0	97.7	91.5	614369;600652
NDRG1	128.4	99.9	98.8	601455

NEFL	164.6	99.7	98.1	607684;607734
NGF	257.6	100.0	100.0	608654
NTRK1	130.6	99.7	97.7	256800
PDK3	105.1	96.4	94.3	300905
PHYH	74.6	97.5	90.8	266500
PLEKHG5	86.9	96.2	89.0	611067;615376
PMP22	111.2	96.7	91.9	118300;118220;145900
PRPS1	149.5	100.0	100.0	311070
PRX	115.6	99.8	98.3	145900;614895
PSAP	114.4	99.9	99.0	611722
RAB7A	157.9	100.0	100.0	600882
REEP1	78.3	76.3	75.7	610250;614751
RETREG1	126.1	95.6	90.1	613115
SBF1	107.9	98.5	96.5	615284
SBF2	117.0	99.6	96.8	604563
SCN10A	165.3	100.0	99.5	615551
SCN11A	138.1	99.2	97.6	615548
SCN9A	146.5	98.5	97.0	243000;133020
SEPT9	118.7	99.7	96.8	162100
SETX	163.2	99.9	99.1	606002;602433
SH3TC2	121.3	100.0	99.7	613353;601596
SLC12A6	141.8	100.0	99.9	218000
SLC25A46	205.7	95.9	87.3	616505
SLC52A2	177.6	100.0	100.0	614707
SLC52A3	119.6	100.0	100.0	211530
SLC5A7	117.1	100.0	99.9	158580
SMN1	112.7	99.8	96.5	271150;253400;253550;253300
SOX10	65.8	98.2	91.3	611584
SPTLC1	115.5	99.0	93.9	162400
SPTLC2	160.2	100.0	100.0	613640

SURF1	96.2	88.3	88.3	616684;256000
SYT2	101.3	100.0	99.0	616040
TDP1	122.9	98.7	95.3	607250
TFG	121.5	93.9	90.7	604484
TRIM2	157.7	93.6	91.4	615490
TRPV4	172.4	99.5	98.7	606071
TTR	152.3	94.6	94.6	105210
TWNK	178.8	100.0	100.0	616138
VCP	144.8	99.9	99.5	167320
WNK1	167.7	99.9	99.5	201300
YARS	122.4	100.0	100.0	608323

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