

# WES HEREDITARY NEUROLOGICAL PAIN DISORDERS<sup>1</sup> DG

## 3.4

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ATL1	153.3	100.0	100.0	182600;613708
ATL3	165.6	100.0	100.0	615632
CABIN1	194.3	100.0	100.0	-
CACNA1A	198.4	100.0	100.0	-
CLTCL1	182.9	100.0	100.0	-
COL6A5	191.0	100.0	100.0	-
COMP	204.9	100.0	100.0	619161
COQ6	189.0	100.0	100.0	614650
DNM1L	144.0	100.0	100.0	614388
DNMT1	188.7	100.0	99.7	614116
DYNC1H1	171.1	100.0	100.0	158600;614228
ELP1	154.9	100.0	100.0	223900
FAAH	172.1	100.0	100.0	606581
FBLN5	144.3	91.8	91.8	No OMIM phenotype
FBN2	164.9	100.0	100.0	No OMIM phenotype
FLVCR1	184.1	100.0	100.0	609033
GLA	161.9	91.3	91.3	301500
HCN1	195.3	98.5	98.5	615871;618482
HCN2	151.8	94.1	91.3	-
HCN3	281.7	100.0	100.0	-
HSPB1	241.3	100.0	100.0	606595;608634
KCNQ3	191.5	100.0	100.0	No OMIM phenotype

KIF1A	185.8	98.0	98.0	614213;610357
LIFR	167.0	100.0	100.0	601559
LZTR1	182.8	100.0	100.0	615670
MME	140.3	98.0	98.0	617017;617018
MPZ	199.3	100.0	100.0	607677;118200;607791;607736
NAGLU	213.5	100.0	100.0	616491;252920
NGF	186.5	100.0	100.0	608654
NMNAT2	154.8	100.0	100.0	-
NTRK1	217.1	100.0	100.0	256800
PIEZO2	173.1	100.0	100.0	617146
PMP22	194.3	100.0	100.0	118220;118300;145900;162500;180800;139393
PRDM12	189.2	96.2	94.0	616488
RAB7A	152.0	100.0	100.0	600882
RETREG1	160.5	100.0	100.0	613115
SCN10A	180.9	100.0	100.0	615551
SCN11A	168.7	100.0	100.0	615548;615552
SCN1B	185.2	100.0	100.0	604233;615377;612838;617350
SCN2B	150.5	100.0	100.0	615378
SCN3A	192.0	100.0	100.0	617935;617938
SCN3B	159.4	100.0	100.0	613120
SCN4B	167.6	100.0	100.0	611819
SCN7A	164.0	100.0	100.0	-
SCN8A	183.6	100.0	100.0	614306;614558;618364;617080
SCN9A	179.1	100.0	100.0	133020;167400;243000
SEPTIN9	227.8	100.0	100.0	162100
SMARCB1	234.5	100.0	100.0	162091
SPTLC1	148.5	100.0	100.0	162400
SPTLC2	150.3	100.0	100.0	613640
TECPR2	179.6	100.0	100.0	No OMIM phenotype
TOR1A	172.1	92.9	91.5	No OMIM phenotype

TRPA1	151.0	100.0	100.0	615040
TRPM8	167.5	100.0	100.0	-
TRPV1	179.4	100.0	100.0	-
TRPV3	174.0	97.1	97.1	614594;616400
TRPV4	193.7	100.0	100.0	606071;600175;617383
TTR	153.8	94.6	94.6	115430;105210
WNK1	180.6	100.0	100.0	201300
ZFHX2	214.8	100.0	100.0	147430

*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.*

*TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.*

*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 no value for coverage are non protein coding genes.*

*Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.*

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