

WES PAINFUL PERIPHERAL NEUROPATHIES¹ DG 3.5

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
GLA	95.1	90.9	90.9	301500
SCN10A	127.9	100.0	100.0	Episodic pain syndrome 615551
SCN11A	136.5	100.0	99.9	615552
SCN1B	110.6	100.0	100.0	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350
SCN2B	105.9	100.0	100.0	'-
SCN3A	160.9	100.0	100.0	-
SCN3B	113.4	100.0	100.0	-
SCN4B	118.2	100.0	100.0	-
SCN7A	159.3	100.0	100.0	-
SCN8A	131.2	100.0	100.0	No OMIM phenotype
SCN9A	157.6	100.0	99.9	133020;167400
TTR	142.6	90.7	90.7	105200;176300

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.

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.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors