

PAINLESS PERIPHERAL NEUROPATHIES PANEL¹ DG 3.8.1

(2 GENES)

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
SCN11A	100.0%	99.9%	99.9%	98.1%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	100.0%	99.9%	100.0%	98.9%	Erythralgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000

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 X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using Twist X2 chemistry.

Twist X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using Twist X2 chemistry.

srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.

srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.

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 : March 17th, 2023.

This list is accurate for panel version DG 3.8.1

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