PAINFUL PERIPHERAL NEUROPATHIES GENE PANEL DG 3.5.0 (12 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
GLA	91%	91%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
SCN10A	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	100%	100%	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838
SCN2B	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3A	100%	100%	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN3B	100%	100%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	100%	100%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN7A	100%	100%	
SCN8A	100%	100%	?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 Developmental and epileptic encephalopathy 13, 614558
SCN9A	100%	100%	Erythermalgia, primary, 133020 Insensitivity to pain, congenital, 243000 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Neuropathy, hereditary sensory and autonomic, type IID, 243000

TTR	91%	91%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430

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.
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TWIST X2 is the chemistry used for WES analysis.
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 coverage denoting NC 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 : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors