SMALL FIBRE NEUROPATHY PANEL[1] DG-5.0.0 (12 GENES)

Gene	***Twist X2 covered 10x***	***Twist X2 covered 20x***	***srWGS covered 10x***	***srWGS covered 15x***	***srWGS covered 20x***	***Associated Phenotype description and OMIM disease ID***
GLA	91.4%	91.4%	98.6%	90.2%	70.6%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
SCN10A	100%	100%	100%	100%	99.5%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	100%	100%	99.8%	99.3%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	100%	100%	100%	99.9%	99.3%	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%	100%	100%	99.6%	Atrial fibrillation, familial, 14, 615378
SCN3A	100%	100%	100%	100%	99.8%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN3B	90.7%	89.7%	100%	100%	98.5%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120

SCN4B	100%	100%	100%	99.9%	98%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN7A	100%	100%	100%	100%	99.9%	
SCN8A	100%	100%	100%	100%	99.6%	?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%	100%	99.9%	99.4%	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	100%	100%	100%	100%	99.9%	Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680

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 mapped against GRCh38.
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 mapped against GRCh38.
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srWGS 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 : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors