SMALL FIBRE NEUROPATHY PANEL[1] DG-4.0.0 (12 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
GLA	91.4%	91.4%	98.4%	73.6%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
SCN10A	100.0%	100.0%	100.0%	98.8%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100.0%	99.9%	99.9%	97.2%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	100.0%	99.9%	100.0%	98.1%	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	100.0%	100.0%	100.0%	98.5%	Atrial fibrillation, familial, 14, 615378

SCN3A	100.0%	100.0%	100.0%	98.2%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN3B	100.0%	100.0%	100.0%	99.0%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4B	100.0%	100.0%	100.0%	98.0%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN7A	100.0%	100.0%	100.0%	98.3%	
SCN8A	100.0%	100.0%	100.0%	98.6%	?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	100.0%	99.9%	100.0%	97.5%	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	100.0%	100.0%	100.0%	99.7%	Amyloidosis, hereditary, transthyretin-related, 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.
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 4.0.0