NEUROLOGICAL PAIN DISORDERS PANEL[1] DG 3.8.1 (62 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ATL1	100.0%	100.0%	100.0%	99.0%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATL3	100.0%	100.0%	100.0%	99.0%	Neuropathy, hereditary sensory, type IF, 615632
CABIN1	100.0%	100.0%	100.0%	99.6%	
CACNA1A	100.0%	100.0%	100.0%	99.2%	Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Developmental and epileptic encephalopathy 42, 617106;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500

CACNA1H	100.0%	100.0%	100.0%	99.4%	{Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CLTCL1	100.0%	100.0%	100.0%	99.8%	
COL6A5	100.0%	99.9%	100.0%	99.2%	
COMP	100.0%	100.0%	100.0%	99.7%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
COQ6	100.0%	100.0%	100.0%	99.5%	Coenzyme Q10 deficiency, primary, 6, 614650
DNM1L	100.0%	100.0%	100.0%	99.5%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT1	99.9%	99.1%	100.0%	99.7%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121

DYNC1H1	100.0%	100.0%	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563
ELP1	100.0%	100.0%	100.0%	99.6%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900
FAAH	100.0%	100.0%	100.0%	99.7%	{Drug addiction, susceptibility to}, 606581
FBLN5	91.8%	91.8%	100.0%	99.6%	Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;Neuropathy, hereditary, with or without age-related macular degeneration, 608895;?Cutis laxa, autosomal dominant 2, 614434
FBN2	100.0%	100.0%	100.0%	99.7%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

FLVCR1	100.0%	100.0%	100.0%	99.7%	Ataxia, posterior column, with retinitis pigmentosa, 609033
GLA	90.9%	90.9%	98.8%	74.8%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
HCN1	99.9%	99.7%	100.0%	99.2%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	94.4%	92.1%	97.3%	89.7%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100.0%	100.0%	100.0%	99.8%	
HSPB1	100.0%	100.0%	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634
KCNQ3	100.0%	100.0%	100.0%	99.3%	Seizures, benign neonatal, 2, 121201

KIF1A	100.0%	100.0%	100.0%	99.9%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 610357
LIFR	100.0%	100.0%	100.0%	99.0%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LZTR1	100.0%	100.0%	100.0%	99.8%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MME	97.6%	97.4%	100.0%	98.9%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MPZ	100.0%	100.0%	100.0%	99.6%	Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1B, 118200;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, dominant intermediate D, 607791;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, type 2J, 607736

NAGLU	100.0%	100.0%	100.0%	99.9%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NGF	100.0%	100.0%	100.0%	99.8%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NMNAT2	100.0%	100.0%	100.0%	98.9%	
NTRK1	100.0%	100.0%	100.0%	99.6%	Insensitivity to pain, congenital, with anhidrosis, 256800
PIEZO2	100.0%	100.0%	100.0%	99.5%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PMP22	100.0%	100.0%	100.0%	98.7%	Charcot-Marie-Tooth disease, type 1A, 118220;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, type 1E, 118300;?Neuropathy, inflammatory demyelinating, 139393;Neuropathy, recurrent, with pressure palsies, 162500;Dejerine-Sottas disease, 145900

PRDM12	95.7%	92.4%	100.0%	98.9%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
RAB7A	100.0%	100.0%	100.0%	99.8%	Charcot-Marie-Tooth disease, type 2B, 600882
RETREG1	100.0%	100.0%	100.0%	99.2%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SCN10A	100.0%	100.0%	100.0%	99.4%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100.0%	99.9%	99.9%	98.1%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	100.0%	100.0%	100.0%	99.7%	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%	99.5%	Atrial fibrillation, familial, 14, 615378
SCN3A	100.0%	100.0%	100.0%	99.0%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938

SCN3B	100.0%	100.0%	100.0%	99.6%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4B	100.0%	100.0%	100.0%	99.0%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN7A	100.0%	100.0%	100.0%	99.4%	
SCN8A	100.0%	100.0%	100.0%	99.3%	?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%	98.9%	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
SEPTIN9	100.0%	100.0%	100.0%	99.3%	Amyotrophy, hereditary neuralgic, 162100

SMARCB1	100.0%	100.0%	100.0%	99.9%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SPTLC1	100.0%	100.0%	100.0%	99.5%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100.0%	100.0%	100.0%	99.5%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
TECPR2	100.0%	100.0%	100.0%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TOR1A	91.2%	90.6%	100.0%	99.3%	Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100;{Dystonia-1, modifier of},
TRPA1	100.0%	100.0%	100.0%	99.3%	?Episodic pain syndrome, familial, 1, 615040
TRPM7	100.0%	100.0%	100.0%	99.2%	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500

TRPM8	100.0%	100.0%	100.0%	99.6%	
TRPV1	100.0%	100.0%	100.0%	99.5%	
TRPV3	100.0%	100.0%	100.0%	99.7%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594
TRPV4	100.0%	100.0%	100.0%	99.7%	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;SED, Maroteaux type, 184095;Metatropic dysplasia, 156530;Scapuloperoneal spinal muscular atrophy, 181405;Hereditary motor and sensory neuropathy, type IIc, 606071;?Avascular necrosis of femoral head, primary, 2, 617383;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500

TTR	90.7%	90.7%	100.0%	99.8%	Amyloidosis, hereditary, transthyretin-related, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
WNK1	100.0%	100.0%	100.0%	99.4%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
ZFHX2	100.0%	100.0%	100.0%	99.6%	?Marsili syndrome, 147430

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