NEUROLOGICAL PAIN DISORDERS PANEL[1] DG-5.1.0 (62 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*
ATL1	100%	100%	100%	100%	99.7%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATL3	100%	100%	100%	100%	99.7%	Neuropathy, hereditary sensory, type IF, 615632
CABIN1	100%	100%	100%	100%	99.5%	
CACNA1A	100%	100%	100%	99.9%	99.1%	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	99.6%	99%	100%	99.9%	99%	{Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CLTCL1	100%	100%	100%	100%	99.6%	
COL6A5	100%	100%	100%	100%	99.8%	

COMP	96.8%	96.3%	100%	99.9%	99.3%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
COQ6	100%	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 6, 614650
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT1	100%	99.3%	100%	100%	99.4%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DYNC1H1	99.3%	99.3%	100%	99.9%	99.4%	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%	100%	100%	100%	99.8%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900
FAAH	100%	100%	100%	100%	99.6%	{Drug addiction, susceptibility to}, 606581

FBLN5	92.8%	92.8%	100%	99.9%	99.4%	Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434
FBN2	99.2%	99.2%	100%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060
GLA	91.4%	91.4%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
HCN1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	95.1%	93.5%	99.3%	97.5%	93%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100%	100%	100%	99.9%	99.1%	

HSPB1	100%	100%	100%	100%	99.1%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634
KCNQ3	100%	100%	100%	100%	99.6%	Seizures, benign neonatal, 2, 121201
KIF1A	100%	100%	100%	100%	99.3%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
LIFR	100%	100%	100%	100%	99.7%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MME	97.7%	97.7%	100%	100%	99.7%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017

MPZ	100%	100%	100%	100%	98.9%	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%	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NGF	100%	100%	100%	100%	98.9%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NMNAT2	100%	100%	100%	100%	99.7%	
NTRK1	100%	100%	100%	99.9%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800
PIEZO2	100%	100%	100%	100%	99.7%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700

PMP22	100%	100%	100%	100%	99.7%	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	97.1%	92.6%	100%	99.9%	98.8%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
RAB7A	89.2%	89.2%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 2B, 600882
RETREG1	91.3%	91.3%	100%	99.9%	99.5%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SCN10A	100%	100%	100%	100%	99.8%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	100%	100%	99.8%	99.4%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	100%	100%	100%	100%	99.5%	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%	99.9%	99%	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%	89.7%	100%	100%	99.7%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4B	100%	100%	100%	100%	99.2%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN7A	100%	100%	100%	99.9%	99.8%	
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%	100%	99.5%	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
SEPTIN9	100%	100%	100%	99.9%	98.9%	Amyotrophy, hereditary neuralgic, 162100

SMARCB1	95.2%	95.2%	100%	100%	99.7%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SPTLC1	84.4%	83.8%	100%	100%	99.7%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	89%	89%	100%	100%	99.7%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
TECPR2	100%	100%	100%	99.9%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TOR1A	93.1%	90.7%	100%	99.9%	99.4%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TRPA1	100%	100%	100%	100%	99.8%	?Episodic pain syndrome, familial, 1, 615040
TRPM7	100%	100%	100%	100%	99.8%	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500
TRPM8	100%	100%	100%	100%	99.7%	
TRPV1	100%	100%	100%	100%	99.3%	
TRPV3	96.6%	96.6%	100%	100%	99.3%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594

TRPV4	100%	100%	100%	99.9%	99.1%	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	100%	100%	100%	100%	99.5%	Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
WNK1	100%	100%	100%	99.9%	99%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
ZFHX2	100%	100%	100%	100%	99.4%	?Marsili syndrome, 147430

Gene symbols used follow HGNC 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.
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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 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors