NEUROLOGICAL PAIN DISORDERS PANEL[1] DG-5.2.0 (62 GENES)

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.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ATL1	100% | 100%	97% | 97%	99% | 99%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATL3	100% | 100%	97% | 97%	99% | 99%	Neuropathy, hereditary sensory, type IF, 615632
CABIN1	100% | 100%	97% | 97%	99% | 99%	
CACNA1A	99% | 99%	95% | 95%	98% | 98%	Developmental and epileptic encephalopathy 42A, 617106;Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500;Developmental and epileptic encephalopathy 42B, 621692
CACNA1H	99% | 98%	94% | 94%	97% | 97%	{Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CLTCL1	100% | 100%	97% | 97%	99% | 99%	
COL6A5	100% | 100%	97% | 97%	99% | 99%	
COMP	96% | 96%	96% | 96%	98% | 98%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
COQ6	100% | 100%	97% | 97%	99% | 99%	Coenzyme Q10 deficiency, primary, 6, 614650
DNM1L	100% | 100%	98% | 98%	99% | 99%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT1	99% | 99%	96% | 96%	99% | 99%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DYNC1H1	99% | 99%	97% | 97%	99% | 99%	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%	98% | 98%	100% | 100%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900

FAAH	99% | 99%	96% | 96%	98% | 98%	{Drug addiction, susceptibility to}, 606581
FBLN5	93% | 93%	96% | 96%	99% | 99%	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% | 99%	98% | 98%	99% | 99%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FLVCR1	95% | 95%	97% | 97%	99% | 99%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060
GLA	91% | 91%	99% | 99%	100% | 100%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
HCN1	100% | 100%	96% | 96%	99% | 99%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	93% | 92%	88% | 88%	97% | 96%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100% | 100%	97% | 97%	99% | 99%	
HSPB1	100% | 100%	96% | 96%	97% | 97%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634
KCNQ3	100% | 100%	96% | 96%	99% | 99%	Seizures, benign neonatal, 2, 121201
KIF1A	100% | 100%	96% | 96%	98% | 98%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
LIFR	100% | 100%	98% | 98%	99% | 99%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LZTR1	100% | 100%	97% | 97%	99% | 99%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MME	98% | 98%	98% | 98%	100% | 100%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MPZ	100% | 100%	96% | 96%	98% | 98%	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%	97% | 97%	97% | 97%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NGF	100% | 100%	96% | 96%	100% | 100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NMNAT2	100% | 100%	97% | 97%	99% | 99%	
NTRK1	100% | 100%	94% | 94%	99% | 99%	Insensitivity to pain, congenital, with anhidrosis, 256800

PIEZO2	100% | 100%	97% | 97%	99% | 99%	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%	97% | 97%	99% | 99%	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	94% | 93%	95% | 95%	98% | 98%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
RAB7A	88% | 88%	96% | 96%	99% | 99%	Charcot-Marie-Tooth disease, type 2B, 600882
RETREG1	91% | 91%	97% | 97%	99% | 99%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SCN10A	100% | 100%	97% | 97%	99% | 99%	Episodic pain syndrome, familial, 2, 615551
SCN11A	99% | 99%	96% | 96%	99% | 99%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	99% | 99%	96% | 96%	99% | 99%	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%	97% | 97%	99% | 99%	Atrial fibrillation, familial, 14, 615378
SCN3A	100% | 100%	98% | 98%	100% | 100%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN3B	91% | 91%	96% | 96%	99% | 99%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4B	100% | 100%	96% | 96%	99% | 99%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN7A	100% | 100%	98% | 98%	99% | 99%	
SCN8A	100% | 100%	96% | 96%	99% | 99%	?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%	98% | 98%	99% | 99%	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	98% | 98%	92% | 92%	98% | 98%	Amyotrophy, hereditary neuralgic, 162100
SMARCB1	96% | 96%	97% | 97%	99% | 99%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SPTLC1	75% | 75%	98% | 98%	99% | 99%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	90% | 90%	97% | 97%	99% | 99%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
TECPR2	100% | 100%	96% | 96%	99% | 99%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031

TOR1A	91% | 91%	97% | 97%	99% | 99%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TRPA1	100% | 100%	97% | 97%	100% | 100%	?Episodic pain syndrome, familial, 1, 615040
TRPM7	100% | 100%	98% | 98%	99% | 99%	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500
TRPM8	100% | 100%	97% | 97%	99% | 99%	
TRPV1	100% | 100%	95% | 95%	99% | 99%	
TRPV3	96% | 96%	96% | 96%	98% | 98%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594
TRPV4	100% | 100%	95% | 95%	99% | 99%	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%	98% | 98%	100% | 100%	Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
WNK1	100% | 100%	96% | 96%	99% | 99%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
ZFHX2	100% | 100%	97% | 97%	99% | 99%	?Marsili syndrome, 147430