HNPD GENE PANEL DG 3.5.0 (251 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
ATL1	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	100%	100%	Neuropathy, hereditary sensory, type IF, 615632
CABIN1	100%	100%	No OMIM disease ID
CACNA1A	100%	100%	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%	100%	Hyperaldosteronism, familial, type IV, 617027
CLTCL1	100%	100%	No OMIM disease ID
COL6A5	100%	100%	No OMIM disease ID
COMP	100%	100%	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400
COQ6	100%	100%	Coenzyme Q10 deficiency, primary, 6, 614650
DNM1L	100%	100%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT1	100%	99%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DYNC1H1	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Intellectual developmental disorder, autosomal dominant 13, 614563
ELP1	100%	100%	Dysautonomia, familial, 223900
FAAH	100%	100%	No OMIM disease ID
FBLN5	92%	92%	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%	100%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FLVCR1	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
GLA	91%	91%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
HCN1	100%	100%	Developmental and epileptic encephalopathy 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	94%	92%	Febrile seizures, familial, 2, 602477 Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100%	100%	No OMIM disease ID
HSPB1	100%	100%	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
KCNQ3	100%	100%	Seizures, benign neonatal, 2, 121201
KIF1A	100%	100%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
LIFR	100%	100%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LZTR1	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MME	98%	97%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MPZ	100%	100%	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%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NGF	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NMNAT2	100%	100%	No OMIM disease ID
NTRK1	100%	100%	Insensitivity to pain, congenital, with anhidrosis, 256800
PIEZO2	100%	100%	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%	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	96%	92%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
RAB7A	100%	100%	Charcot-Marie-Tooth disease, type 2B, 600882
RETREG1	100%	100%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SCN10A	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	100%	100%	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%	Atrial fibrillation, familial, 14, 615378
SCN3A	100%	100%	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN3B	100%	100%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	100%	100%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN7A	100%	100%	No OMIM disease ID
SCN8A	100%	100%	?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%	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%	Amyotrophy, hereditary neuralgic, 162100

SMARCB1	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SPTLC1	100%	100%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
TECPR2	100%	100%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TOR1A	91%	91%	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100
TRPA1	100%	100%	?Episodic pain syndrome, familial, 1, 615040
TRPM7	100%	100%	No OMIM disease ID
TRPM8	100%	100%	No OMIM disease ID
TRPV1	100%	100%	No OMIM disease ID
TRPV3	100%	100%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome 1, 614594
TRPV4	100%	100%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Digital arthropathy-brachydactyly, familial, 606835 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 Neuronopathy, distal hereditary motor, type VIII, 600175 Parastremmatic dwarfism, 168400 Brachyolmia type 3, 113500
TTR	91%	91%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
WNK1	100%	100%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
ZFHX2	100%	100%	?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.
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TWIST X2 is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 28th , 2022.

This list is accurate for panel version DG 3.5.0

Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors