HNPD GENE PANEL DG 2.18 (50 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ATL1	100%	99,70%	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	99,80%	98,30%	100%	100%	Neuropathy, hereditary sensory, type IF, 615632
CABIN1	100%	99,60%	100%	99,90%	No OMIM disease ID
CLTCL1	98,60%	98,20%	100%	100%	No OMIM disease ID
COL6A5	99,90%	99,50%	100%	100%	No OMIM disease ID
COQ6	99,90%	98,40%	100%	100%	Coenzyme Q10 deficiency, primary, 6, 614650
DNM1L	99,90%	98,50%	100%	100%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNMT1	99,20%	99,00%	99,70%	99,20%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DYNC1H1	99,90%	99,40%	100%	100%	Mental retardation, autosomal dominant 13, 614563 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
ELP1	99,80%	99,00%	100%	100%	Dysautonomia, familial, 223900
FAAH	93,20%	90,00%	100%	99,90%	No OMIM disease ID
FLVCR1	100%	98,90%	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
GLA	99,80%	96,60%	100%	100%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
HCN1	100%	99,70%	100%	100%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
HCN2	59,20%	49,50%	84,10%	77,30%	No OMIM disease ID
HCN3	99,90%	98,50%	100%	100%	No OMIM disease ID
HSPB1	98,80%	91,60%	100%	100%	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
KIF1A	99,40%	97,10%	100%	100%	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
LIFR	99,70%	98,00%	100%	100%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559

LZTR1	100%	99,90%	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MPZ	100%	100%	100%	100%	Charcot-Marie-Tooth disease, type 2J, 607736 Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677
NAGLU	92,90%	89,90%	99,90%	99,20%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NGF	100%	100%	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NMNAT2	99,90%	98,90%	100%	100%	No OMIM disease ID
NTRK1	99,80%	98,20%	100%	100%	Insensitivity to pain, congenital, with anhidrosis, 256800
PIEZO2	100%	99,50%	100%	100%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PRDM12	90,80%	88,00%	93,40%	91,70%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
RAB7A	100%	99,90%	100%	100%	Charcot-Marie-Tooth disease, type 2B, 600882
RETREG1	98,80%	95,10%	100%	100%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SCN10A	100%	99,70%	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN11A	99,80%	98,30%	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1B	98,00%	96,40%	99,80%	99,30%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2B	100%	100%	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3A	99,90%	99,30%	100%	100%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN3B	100%	100%	100%	100%	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120
SCN4B	100%	99,60%	100%	100%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN7A	98,30%	93,20%	100%	100%	No OMIM disease ID

SCN8A	100%	99,80%	100%	100%	Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364 Epileptic encephalopathy, early infantile, 13, 614558
SCN9A	99,30%	97,80%	100%	100%	Small fiber neuropathy, 133020 HSAN2D, autosomal recessive, 243000 Paroxysmal extreme pain disorder, 167400 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SEPT9	100%	99,90%	100%	100%	Amyotrophy, hereditary neuralgic, 162100
SMARCB1	100%	100%	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SPTLC1	99,20%	95,40%	100%	100%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100%	100%	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
TRPA1	96,10%	89,80%	100%	100%	?Episodic pain syndrome, familial, 1, 615040
TRPM8	99,80%	98,80%	100%	100%	No OMIM disease ID
TRPV1	100%	99,60%	100%	100%	No OMIM disease ID
TRPV3	99,80%	98,50%	97,10%	97,10%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	100%	99,90%	100%	100%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 Scapuloperoneal spinal muscular atrophy, 181405 Metatropic dysplasia, 156530 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Brachyolmia type 3, 113500 ?Avascular necrosis of femoral head, primary, 2, 617383
TTR	94,60%	94,60%	94,60%	94,60%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
WNK1	99,90%	99,60%	100%	100%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300

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.

Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors