HNPD GENE PANEL DG 3.00 (55 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ATL1	100	99,7	100	100	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	99,8	98,3	100	100	Neuropathy, hereditary sensory, type IF, 615632
CABIN1	100	99,6	100	99,9	No OMIM disease ID
CLTCL1	98,6	98,2	100	100	No OMIM disease ID
COL6A5	99,9	99,5	100	100	No OMIM disease ID
COQ6	99,9	98,4	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
DNM1L	99,9	98,5	100	100	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNMT1	99,2	99	99,7	99,2	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DYNC1H1	99,9	99,4	100	100	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228
ELP1	99,8	99	100	100	Dysautonomia, familial, 223900
FAAH	93,2	90	100	99,9	{Drug addiction, susceptibility to}, 606581
FBLN5	91,8	91,8	91,8	91,8	Macular degeneration, age-related, 3, 608895 ?Cutis laxa, autosomal dominant 2, 614434 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 Cutis laxa, autosomal recessive, type IA, 219100
FLVCR1	100	98,9	100	100	Ataxia, posterior column, with retinitis pigmentosa, 609033
GLA	91,1	88,2	91,3	91,3	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
HCN1	98,5	98,2	98,5	98,5	Generalized epilepsy with febrile seizures plus, type 10, 618482 Developmental and epileptic encephalopathy 24, 615871

HCN2	59,2	49,5	84,1	77,3	No OMIM disease ID
HCN3	99,9	98,5	100	100	No OMIM disease ID
HSPB1	98,8	91,6	100	100	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
KIF1A	97,4	95,2	98	98	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
LIFR	99,7	98	100	100	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MME	99,8	98,7	98	98	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MPZ	87,9	84,1	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	92,9	89,9	99,9	99,2	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	99,9	98,9	100	100	No OMIM disease ID
NTRK1	99,8	98,2	100	100	Insensitivity to pain, congenital, with anhidrosis, 256800
PIEZO2	100	99,5	100	100	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PMP22	100	100	100	100	Dejerine-Sottas disease, 145900 ?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800

					Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1A, 118220
PRDM12	90,8	88	93,4	91,7	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
RAB7A	100	99,9	100	100	Charcot-Marie-Tooth disease, type 2B, 600882
RETREG1	98,8	95,1	100	100	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SCN10A	100	99,6	100	100	Episodic pain syndrome, familial, 2, 615551
SCN11A	99,8	98,3	100	100	Neuropathy, hereditary sensory and autonomic, type VII, 615548 Episodic pain syndrome, familial, 3, 615552
SCN1B	98	96,4	99,8	99,3	Atrial fibrillation, familial, 13, 615377 Developmental and epileptic encephalopathy 52, 617350 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	99,8	99,2	100	100	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN3B	100	100	100	100	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120
SCN4B	100	99,6	100	100	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN7A	98,3	93,3	100	100	No OMIM disease ID
SCN8A	100	99,8	100	100	Seizures, benign familial infantile, 5, 617080 Developmental and epileptic encephalopathy 13, 614558 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364
SCN9A	99,3	97,9	100	100	Neuropathy, hereditary sensory and autonomic, type IID, 243000 Generalized epilepsy with febrile seizures plus, type 7, 613863 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SEPTIN9	100	99,9	100	100	Amyotrophy, hereditary neuralgic, 162100

SMARCB1	100	100	100	100	Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091 Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322
SPTLC1	99,2	95,4	100	100	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100	100	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
TRPA1	96,1	89,8	100	100	?Episodic pain syndrome, familial, 1, 615040
TRPM8	99,8	98,8	100	100	No OMIM disease ID
TRPV1	100	99,6	100	100	No OMIM disease ID
TRPV3	99,8	98,5	97,1	97,1	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	100	99,9	100	100	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 [Sodium serum level QTL 1], 613508 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	94,6	94,6	94,6	94,6	Amyloidosis, hereditary, transthyretin-related, 105210 [Dystransthyretinemic hyperthyroxinemia], 145680 Carpal tunnel syndrome, familial, 115430
WNK1	99,9	99,6	100	100	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
ZFHX2	100	99,6	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.
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 : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors