# HNPD GENE PANEL DG 3.2.0 (58 genes)

Releasedate: 16-09-2021

| *Gene* | *Agilent V5 covered >10x* | *Agilent V5 covered >20x* | *TWIST covered >10x* | *TWIST covered >20x* | *Associated Phenotype Description and OMIM disease ID* |
|---|---|---|---|---|---|
| ATL1 | 99,9 | 99,5 | 100 | 99,8 | Spastic paraplegia 3A, autosomal dominant, 182600<br>Neuropathy, hereditary sensory, type ID, 613708 |
| ATL3 | 99,6 | 97,6 | 100 | 99,9 | Neuropathy, hereditary sensory, type IF, 615632 |
| CABIN1 | 100 | 99,4 | 100 | 99,9 | No OMIM disease ID |
| CACNA1A | 93,1 | 88,4 | 100 | 99,9 | Developmental and epileptic encephalopathy 42, 617106<br>Spinocerebellar ataxia 6, 183086<br>Episodic ataxia, type 2, 108500<br>Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500<br>Migraine, familial hemiplegic, 1, 141500 |
| CLTCL1 | 98,6 | 97,5 | 100 | 100 | No OMIM disease ID |
| COL6A5 | 99,9 | 99,2 | 100 | 100 | No OMIM disease ID |
| COMP | 93,8 | 92,4 | 100 | 100 | Pseudoachondroplasia, 177170<br>Carpal tunnel syndrome 2, 619161<br>Epiphyseal dysplasia, multiple, 1, 132400 |
| COQ6 | 99,9 | 98,5 | 100 | 100 | Coenzyme Q10 deficiency, primary, 6, 614650 |
| DNM1L | 99,6 | 98,3 | 100 | 100 | Optic atrophy 5, 610708<br>Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 |
| DNMT1 | 99,2 | 98,8 | 99,9 | 99,4 | Neuropathy, hereditary sensory, type IE, 614116<br>Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 |
| DYNC1H1 | 99,9 | 99,3 | 100 | 100 | Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600<br>Charcot-Marie-Tooth disease, axonal, type 20, 614228<br>Mental retardation, autosomal dominant 13, 614563 |
| ELP1 | 99,8 | 98,9 | 100 | 100 | Dysautonomia, familial, 223900 |
| FAAH | 94,3 | 90,3 | 100 | 100 | No OMIM disease ID |
| FBLN5 | 91,8 | 91,7 | 91,8 | 91,8 | Cutis laxa, autosomal recessive, type IA, 219100<br>Macular degeneration, age-related, 3, 608895<br>Neuropathy, hereditary, with or without age-related macular degeneration, 608895<br>?Cutis laxa, autosomal dominant 2, 614434 |

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| FLVCR1 | 99,7 | 98,3 | 100 | 99,9 | Ataxia, posterior column, with retinitis pigmentosa, 609033 |
| GLA | 91 | 85,9 | 91,3 | 91,3 | Fabry disease, cardiac variant, 301500<br>Fabry disease, 301500 |
| HCN1 | 98,4 | 98,3 | 98,5 | 98,4 | Developmental and epileptic encephalopathy 24, 615871<br>Generalized epilepsy with febrile seizures plus, type 10, 618482 |
| HCN2 | 59,8 | 47,7 | 84 | 76,9 | Febrile seizures, familial, 2, 602477<br>Generalized epilepsy with febrile seizures plus, type 11, 602477 |
| HCN3 | 99,9 | 98,5 | 100 | 100 | No OMIM disease ID |
| HSPB1 | 99,1 | 92,1 | 100 | 100 | Neuronopathy, distal hereditary motor, type IIB, 608634<br>Charcot-Marie-Tooth disease, axonal, type 2F, 606595 |
| KIF1A | 97,4 | 95,3 | 98 | 98 | NESCAV syndrome, 614255<br>Neuropathy, hereditary sensory, type IIC, 614213<br>Spastic paraplegia 30, autosomal dominant, 610357<br>Spastic paraplegia 30, autosomal recessive, 610357 |
| LIFR | 99,3 | 97,8 | 100 | 99,9 | Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 |
| LZTR1 | 100 | 99,9 | 100 | 100 | Noonan syndrome 2, 605275<br>Noonan syndrome 10, 616564 |
| MME | 99,7 | 98,6 | 98 | 97,9 | ?Spinocerebellar ataxia 43, 617018<br>Charcot-Marie-Tooth disease, axonal, type 2T, 617017 |
| MPZ | 85,6 | 81,9 | 100 | 100 | Charcot-Marie-Tooth disease, type 2I, 607677<br>Dejerine-Sottas disease, 145900<br>Charcot-Marie-Tooth disease, type 1B, 118200<br>Roussy-Levy syndrome, 180800<br>Charcot-Marie-Tooth disease, dominant intermediate D, 607791<br>Hypomyelinating neuropathy, congenital, 2, 618184<br>Charcot-Marie-Tooth disease, type 2J, 607736 |
| NAGLU | 93,8 | 91,7 | 99,9 | 98,7 | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491<br>Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 |
| NGF | 100 | 100 | 100 | 100 | Neuropathy, hereditary sensory and autonomic, type V, 608654 |
| NMNAT2 | 100 | 98,8 | 100 | 100 | No OMIM disease ID |
| NTRK1 | 99,9 | 98,5 | 100 | 100 | Insensitivity to pain, congenital, with anhidrosis, 256800 |
| PIEZO2 | 99,8 | 99,2 | 100 | 100 | Arthrogryposis, distal, type 5, 108145<br>Arthrogryposis, distal, with impaired proprioception and touch, 617146<br>Arthrogryposis, distal, type 3, 114300<br>?Marden-Walker syndrome, 248700 |

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| PMP22 | 100 | 100 | 100 | 100 | Charcot-Marie-Tooth disease, type 1A, 118220<br>Roussy-Levy syndrome, 180800<br>Charcot-Marie-Tooth disease, type 1E, 118300<br>?Neuropathy, inflammatory demyelinating, 139393<br>Neuropathy, recurrent, with pressure palsies, 162500<br>Dejerine-Sottas disease, 145900 |
| PRDM12 | 91,7 | 89,6 | 92,8 | 91 | Neuropathy, hereditary sensory and autonomic, type VIII, 616488 |
| RAB7A | 100 | 100 | 100 | 100 | Charcot-Marie-Tooth disease, type 2B, 600882 |
| RETREG1 | 99,1 | 96,1 | 100 | 100 | Neuropathy, hereditary sensory and autonomic, type IIB, 613115 |
| SCN10A | 99,9 | 98,5 | 100 | 100 | Episodic pain syndrome, familial, 2, 615551 |
| SCN11A | 99,3 | 97,5 | 100 | 100 | Episodic pain syndrome, familial, 3, 615552<br>Neuropathy, hereditary sensory and autonomic, type VII, 615548 |
| SCN1B | 98,2 | 96,3 | 99,7 | 98,9 | Generalized epilepsy with febrile seizures plus, type 1, 604233<br>Developmental and epileptic encephalopathy 52, 617350<br>Cardiac conduction defect, nonspecific, 612838<br>Atrial fibrillation, familial, 13, 615377<br>Brugada syndrome 5, 612838 |
| SCN2B | 100 | 100 | 100 | 100 | Atrial fibrillation, familial, 14, 615378 |
| SCN3A | 99,8 | 99,1 | 100 | 100 | Epilepsy, familial focal, with variable foci 4, 617935<br>Developmental and epileptic encephalopathy 62, 617938 |
| SCN3B | 100 | 100 | 100 | 100 | Atrial fibrillation, familial, 16, 613120<br>Brugada syndrome 7, 613120 |
| SCN4B | 99,9 | 97,1 | 100 | 100 | Atrial fibrillation, familial, 17, 611819<br>Long QT syndrome 10, 611819 |
| SCN7A | 97,7 | 91,2 | 100 | 99,9 | No OMIM disease ID |
| SCN8A | 100 | 99,5 | 100 | 100 | ?Myoclonus, familial, 2, 618364<br>Seizures, benign familial infantile, 5, 617080<br>Cognitive impairment with or without cerebellar ataxia, 614306<br>Developmental and epileptic encephalopathy 13, 614558 |
| SCN9A | 99,1 | 97 | 100 | 100 | Erythermalgia, primary, 133020<br>Insensitivity to pain, congenital, 243000<br>Small fiber neuropathy, 133020<br>Paroxysmal extreme pain disorder, 167400<br>Neuropathy, hereditary sensory and autonomic, type IID, 243000 |
| SEPTIN9 | 100 | 99,5 | 100 | 100 | Amyotrophy, hereditary neuralgic, 162100 |

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| SMARCB1 | 100 | 99,9 | 100 | 100 | Rhabdoid tumors, somatic, 609322<br>Coffin-Siris syndrome 3, 614608 |
| SPTLC1 | 98,7 | 93,7 | 100 | 100 | Neuropathy, hereditary sensory and autonomic, type IA, 162400 |
| SPTLC2 | 100 | 100 | 100 | 99,9 | Neuropathy, hereditary sensory and autonomic, type IC, 613640 |
| TECPR2 | 100 | 100 | 100 | 100 | Spastic paraplegia 49, autosomal recessive, 615031 |
| TOR1A | 91,3 | 91,3 | 91,7 | 91,3 | Arthrogryposis multiplex congenita 5, 618947<br>Dystonia-1, torsion, 128100 |
| TRPA1 | 96,4 | 89,9 | 100 | 99,9 | ?Episodic pain syndrome, familial, 1, 615040 |
| TRPM8 | 99,8 | 98,5 | 100 | 100 | No OMIM disease ID |
| TRPV1 | 99,9 | 99 | 100 | 100 | No OMIM disease ID |
| TRPV3 | 99,8 | 98,6 | 97,1 | 97,1 | ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400<br>Olmsted syndrome 1, 614594 |
| TRPV4 | 100 | 99,9 | 100 | 100 | Spondylometaphyseal dysplasia, Kozlowski type, 184252<br>Digital arthropathy-brachydactyly, familial, 606835<br>SED, Maroteaux type, 184095<br>Metatropic dysplasia, 156530<br>Scapuloperoneal spinal muscular atrophy, 181405<br>Hereditary motor and sensory neuropathy, type IIc, 606071<br>?Avascular necrosis of femoral head, primary, 2, 617383<br>Neuronopathy, distal hereditary motor, type VIII, 600175<br>Parastremmatic dwarfism, 168400<br>Brachyolmia type 3, 113500 |
| TTR | 94,6 | 94,6 | 94,6 | 94,6 | Amyloidosis, hereditary, transthyretin-related, 105210<br>Carpal tunnel syndrome, familial, 115430 |
| WNK1 | 99,8 | 99,3 | 100 | 100 | Neuropathy, hereditary sensory and autonomic, type II, 201300<br>Pseudohypoaldosteronism, type IIC, 614492 |
| ZFHX2 | 99,8 | 99,1 | 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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*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-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 : September 16th , 2021.*
*This list is accurate for panel version DG 3.2.0*
*Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors*