# NEUROPATHIES HMSN GENE PANEL DG 2.12 (78 genes)

| Gene | Median | % covered > 10x | % covered > 20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|
| AARS | 124.4 | 100 | 99 | Charcot-Marie-Tooth disease, axonal, type 2N, 613287<br>Epileptic encephalopathy, early infantile, 29, 616339 |
| ABHD12 | 107 | 97 | 87 | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 |
| AIFM1 | 107 | 100 | 99 | Combined oxidative phosphorylation deficiency 6, 300816<br>Cowchock syndrome, 310490<br>Deafness, X-linked 5, 300614 |
| ATL1 | 160.3 | 99 | 97 | Neuropathy, hereditary sensory, type ID, 613708<br>Spastic paraplegia 3A, autosomal dominant, 182600 |
| ATP7A | 134.1 | 99 | 97 | Menkes disease, 309400<br>Occipital horn syndrome, 304150<br>Spinal muscular atrophy, distal, X-linked 3, 300489 |
| BSCL2 | 113.7 | 100 | 100 | Encephalopathy, progressive, with or without lipodystrophy, 615924<br>Lipodystrophy, congenital generalized, type 2, 269700<br>Neuropathy, distal hereditary motor, type VA, 600794<br>Silver spastic paraplegia syndrome, 270685 |
| C10orf2 | 179 | 100 | 100 | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245<br>Perrault syndrome 5, 616138<br>Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 |
| COX6A1 | 181.3 | 100 | 99 | Charcot-Marie-Tooth disease, recessive intermediate D, 616039 |
| CTDP1 | 105.3 | 86 | 83 | Congenital cataracts, facial dysmorphism, and neuropathy, 604168 |
| DCTN1 | 131.9 | 99 | 98 | Neuropathy, distal hereditary motor, type VIIB, 607641<br>Perry syndrome, 168605<br>{Amyotrophic lateral sclerosis, susceptibility to}, 105400 |
| DHH | 117.7 | 100 | 100 | 46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080<br>46XY sex reversal 7, 233420 |
| DHTKD1 | 141.1 | 99 | 98 | 2-aminoadipic 2-oxoadipic aciduria, 204750<br>?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 |
| DNAJB2 | 102.1 | 100 | 100 | Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 |

| | | | | |
|---|---|---|---|---|
| DNM2 | 127.9 | 97 | 94 | Charcot-Marie-Tooth disease, axonal, type 2M, 606482<br>Charcot-Marie-Tooth disease, dominant intermediate B, 606482<br>Lethal congenital contracture syndrome 5, 615368<br>Myopathy, centronuclear, 160150 |
| DNMT1 | 113.5 | 99 | 98 | Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121<br>Neuropathy, hereditary sensory, type IE, 614116 |
| DYNC1H1 | 180 | 100 | 99 | Charcot-Marie-Tooth disease, axonal, type 20, 614228<br>Mental retardation, autosomal dominant 13, 614563<br>Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 |
| EGR2 | 124.6 | 100 | 100 | Charcot-Marie-Tooth disease, type 1D, 607678<br>Dejerine-Sottas disease, 145900<br>Neuropathy, congenital hypomyelinating, 1, 605253 |
| EXOSC8 | 79.9 | 91 | 76 | Pontocerebellar hypoplasia, type 1C, 616081 |
| FAM134B | 126.1 | 95 | 89 | Neuropathy, hereditary sensory and autonomic, type IIB, 613115 |
| FGD4 | 110.2 | 98 | 96 | Charcot-Marie-Tooth disease, type 4H, 609311 |
| FIG4 | 154.9 | 99 | 98 | Amyotrophic lateral sclerosis 11, 612577<br>Charcot-Marie-Tooth disease, type 4J, 611228<br>Yunis-Varon syndrome, 216340<br>?Polymicrogyria, bilateral temporooccipital, 612691 |
| GAN | 190.2 | 100 | 99 | Giant axonal neuropathy-1, 256850 |
| GARS | 125.1 | 99 | 98 | Charcot-Marie-Tooth disease, type 2D, 601472<br>Neuropathy, distal hereditary motor, type VA, 600794 |
| GDAP1 | 163 | 99 | 96 | Charcot-Marie-Tooth disease, axonal, type 2K, 607831<br>Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706<br>Charcot-Marie-Tooth disease, recessive intermediate, A, 608340<br>Charcot-Marie-Tooth disease, type 4A, 214400 |
| GJB1 | 231.2 | 100 | 99 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 |
| GNB4 | 152.5 | 100 | 99 | Charcot-Marie-Tooth disease, dominant intermediate F, 615185 |
| HINT1 | 60 | 98 | 88 | Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 |
| HOXD10 | 137.8 | 100 | 99 | Charcot-Marie-Tooth disease, foot deformity of, 192950<br>Vertical talus, congenital, 192950 |
| HSPB1 | 39.9 | 93 | 81 | Charcot-Marie-Tooth disease, axonal, type 2F, 606595<br>Neuropathy, distal hereditary motor, type IIB, 608634 |
| HSPB8 | 156.4 | 100 | 100 | Charcot-Marie-Tooth disease, axonal, type 2L, 608673 |

| | | | | |
|---|---|---|---|---|
| | | | | Neuropathy, distal hereditary motor, type IIA, 158590 |
| IGHMBP2 | 107.9 | 99 | 96 | Charcot-Marie-Tooth disease, axonal, type 2S, 616155<br>Neuronopathy, distal hereditary motor, type VI, 604320 |
| IKBKAP | 142.7 | 99 | 98 | Dysautonomia, familial, 223900 |
| INF2 | 88.2 | 93 | 90 | Charcot-Marie-Tooth disease, dominant intermediate E, 614455<br>Glomerulosclerosis, focal segmental, 5, 613237 |
| KARS | 122.7 | 100 | 99 | Deafness, autosomal recessive 89, 613916<br>?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 |
| KIF1A | 114.2 | 99 | 96 | Mental retardation, autosomal dominant 9, 614255<br>Neuropathy, hereditary sensory, type IIC, 614213<br>Spastic paraplegia 30, autosomal recessive, 610357 |
| KIF1B | 154.8 | 100 | 99 | Pheochromocytoma, 171300<br>?Charcot-Marie-Tooth disease, type 2A1, 118210<br>{Neuroblastoma, susceptibility to, 1}, 256700 |
| KIF5A | 136.3 | 100 | 99 | Spastic paraplegia 10, autosomal dominant, 604187 |
| LITAF | 126.6 | 94 | 91 | Charcot-Marie-Tooth disease, type 1C, 601098 |
| LMNA | 89.4 | 98 | 91 | Cardiomyopathy, dilated, 1A, 115200<br>Charcot-Marie-Tooth disease, type 2B1, 605588<br>Emery-Dreifuss muscular dystrophy 2, AD, 181350<br>Emery-Dreifuss muscular dystrophy 3, AR, 616516<br>Heart-hand syndrome, Slovenian type, 610140<br>Hutchinson-Gilford progeria, 176670<br>Lipodystrophy, familial partial, 2, 151660<br>Malouf syndrome, 212112<br>Mandibuloacral dysplasia, 248370<br>Muscular dystrophy, congenital, 613205<br>Muscular dystrophy, limb-girdle, type 1B, 159001<br>Restrictive dermopathy, lethal, 275210 |
| LRSAM1 | 130.5 | 100 | 99 | Charcot-Marie-Toothe disease, axonal, type 2P, 614436 |
| MED25 | 104.1 | 99 | 95 | Basel-Vanagait-Smirin-Yosef syndrome, 616449<br>?Charcot-Marie-Tooth disease, type 2B2, 605589 |
| MFN2 | 150.9 | 100 | 99 | Charcot-Marie-Tooth disease, type 2A2A, 609260<br>Charcot-Marie-Tooth disease, type 2A2B, 617087<br>Hereditary motor and sensory neuropathy VIA, 601152 |

| | | | | |
|---|---|---|---|---|
| MME | 100.6 | 98 | 93 | Charcot-Marie-Tooth disease, axonal, type 2T, 617017<br>?Spinocerebellar ataxia 43, 617018 |
| MPZ | 118.7 | 99 | 97 | Charcot-Marie-Tooth disease, dominant intermediate D, 607791<br>Charcot-Marie-Tooth disease, type 1B, 118200<br>Charcot-Marie-Tooth disease, type 2I, 607677<br>Charcot-Marie-Tooth disease, type 2J, 607736<br>Dejerine-Sottas disease, 145900<br>Neuropathy, congenital hypomyelinating, 605253<br>Roussy-Levy syndrome, 180800 |
| MTMR2 | 106.5 | 100 | 99 | Charcot-Marie-Tooth disease, type 4B1, 601382 |
| NDRG1 | 128.7 | 99 | 98 | Charcot-Marie-Tooth disease, type 4D, 601455 |
| NEFL | 165 | 99 | 98 | Charcot-Marie-Tooth disease, type 1F, 607734<br>Charcot-Marie-Tooth disease, type 2E, 607684 |
| NGF | 258.2 | 100 | 100 | Neuropathy, hereditary sensory and autonomic, type V, 608654 |
| NTRK1 | 130.9 | 99 | 97 | Insensitivity to pain, congenital, with anhidrosis, 256800<br>Medullary thyroid carcinoma, familial, 155240 |
| PHYH | 74.7 | 97 | 90 | Refsum disease, 266500 |
| PLEKHG5 | 87 | 96 | 88 | Charcot-Marie-Tooth disease, recessive intermediate C, 615376<br>Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 |
| PMP22 | 111.4 | 96 | 91 | Charcot-Marie-Tooth disease, type 1A, 118220<br>Charcot-Marie-Tooth disease, type 1E, 118300<br>Dejerine-Sottas disease, 145900<br>Neuropathy, inflammatory demyelinating, 139393<br>Neuropathy, recurrent, with pressure palsies, 162500<br>Roussy-Levy syndrome, 180800 |
| PRPS1 | 150.5 | 100 | 100 | Arts syndrome, 301835<br>Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070<br>Deafness, X-linked 1, 304500<br>Gout, PRPS-related, 300661<br>Phosphoribosylpyrophosphate synthetase superactivity, 300661 |
| PRX | 115.8 | 99 | 98 | Charcot-Marie-Tooth disease, type 4F, 614895<br>Dejerine-Sottas disease, 145900 |

| | | | | |
|---|---|---|---|---|
| PSAP | 114.3 | 99 | 99 | Combined SAP deficiency, 611721<br>Gaucher disease, atypical, 610539<br>Krabbe disease, atypical, 611722<br>Metachromatic leukodystrophy due to SAP-b deficiency, 249900 |
| RAB7A | 158.2 | 100 | 100 | Charcot-Marie-Tooth disease,type 2B, 600882 |
| SBF2 | 116.9 | 99 | 96 | Charcot-Marie-Tooth disease, type 4B2, 604563 |
| SCN10A | 165.5 | 100 | 99 | Episodic pain syndrome, familial, 2, 615551 |
| SCN11A | 138 | 99 | 97 | Episodic pain syndrome, familial, 3, 615552<br>Neuropathy, hereditary sensory and autonomic, type VII, 615548 |
| SCN9A | 148.3 | 98 | 97 | Epilepsy, generalized, with febrile seizures plus, type 7, 613863<br>Erythermalgia, primary, 133020<br>Febrile seizures, familial, 3B, 613863<br>HSAN2D, autosomal recessive, 243000<br>Insensitivity to pain, congenital, 243000<br>Paroxysmal extreme pain disorder, 167400,<br>Small fiber neuropathy, 133020<br>{Dravet syndrome, modifier of}, 607208 |
| SEPT9 | 118.8 | 99 | 96 | Amyotrophy, hereditary neuralgic, 162100<br>Leukemia, acute myeloid, therapy-related<br>Ovarian carcinoma |
| SH3TC2 | 122.8 | 100 | 99 | Charcot-Marie-Tooth disease, type 4C, 601596<br>Mononeuropathy of the median nerve, mild, 613353 |
| SLC12A6 | 141.8 | 100 | 99 | Agenesis of the corpus callosum with peripheral neuropathy, 218000 |
| SLC52A2 | 177.8 | 100 | 100 | Brown-Vialetto-Van Laere syndrome 2, 614707 |
| SLC52A3 | 119.7 | 100 | 100 | Brown-Vialetto-Van Laere syndrome 1, 211530<br>Fazio-Londe disease, 211500 |
| SLC5A7 | 117.2 | 100 | 99 | Neuronopathy, distal hereditary motor, type VIIA, 158580 |
| SMN1 | 116.3 | 99 | 96 | Spinal muscular atrophy-1, 253300<br>Spinal muscular atrophy-2, 253550<br>Spinal muscular atrophy-3, 253400<br>Spinal muscular atrophy-4, 271150 |
| SOX10 | 65.8 | 98 | 91 | PCWH syndrome, 609136<br>Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584<br>Waardenburg syndrome, type 4C, 613266 |
| SPTLC1 | 115.4 | 98 | 94 | Neuropathy, hereditary sensory and autonomic, type IA, 162400 |

| | | | | |
|---|---|---|---|---|
| SPTLC2 | 160.3 | 100 | 100 | Neuropathy, hereditary sensory and autonomic, type IC, 613640 |
| SYT2 | 101.3 | 100 | 99 | Myasthenic syndrome, congenital, 7, presynaptic, 616040 |
| TDP1 | 123 | 98 | 95 | Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250 |
| TFG | 125.3 | 96 | 93 | Hereditary motor and sensory neuropathy, Okinawa type, 604484<br>?Spastic paraplegia 57, autosomal recessive, 615658 |
| TRPV4 | 172.7 | 99 | 98 | Brachyolmia type 3, 113500<br>Digital arthropathy-brachydactyly, familial, 606835<br>Hereditary motor and sensory neuropathy, type IIc, 606071<br>Metatropic dysplasia, 156530<br>Parastremmatic dwarfism, 168400<br>Scapuloperoneal spinal muscular atrophy, 181405<br>SED, Maroteaux type, 184095<br>Spinal muscular atrophy, distal, congenital nonprogressive, 600175<br>Spondylometaphyseal dysplasia, Kozlowski type, 184252<br>[Sodium serum level QTL 1], 613508 |
| TTR | 159.7 | 100 | 100 | Amyloidosis, hereditary, transthyretin-related, 105210<br>Carpal tunnel syndrome, familial, 115430<br>[Dystransthyretinemic hyperthyroxinemia], 145680 |
| VCP | 144.9 | 99 | 99 | Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954<br>Charcot-Marie-Tooth disease, type 2Y, 616687<br>Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 |
| WNK1 | 167.2 | 99 | 99 | Neuropathy, hereditary sensory and autonomic, type II, 201300<br>Pseudohypoaldosteronism, type IIC, 614492 |
| YARS | 122.6 | 100 | 100 | Charcot-Marie-Tooth disease, dominant intermediate C, 608323 |

*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.*
*Median Coverage describes the average number of reads seen across 50 exomes.*
*% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.*
*OMIM release used for OMIM disease identifiers and descriptions : April 14th, 2017.*
*This list is accurate for panel version DG 2.12*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*