NEUROPATHIES GENE PANEL DG 3.4.0 (230 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100,0%	100,0%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100,0%	100,0%	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287 ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 Trichothiodystrophy 8, nonphotosensitive, 619691
ABCA1	100,0%	100,0%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCD1	100,0%	100,0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	100,0%	100,0%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACOX1	100,0%	100,0%	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ADPRS	100,0%	100,0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	100,0%	100,0%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGRN	100,0%	100,0%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGTPBP1	100,0%	100,0%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AHNAK2	97,7%	97,6%	No OMIM Disease ID
AIFM1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
AMACR	100,0%	100,0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
APTX	100,0%	100,0%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARHGEF10	100,0%	100,0%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF28	100,0%	100,0%	No OMIM Disease ID
ARSA	100,0%	100,0%	Metachromatic leukodystrophy, 250100

ATAD3A	100,0%	100,0%	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATL1	100,0%	100,0%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	100,0%	100,0%	Neuropathy, hereditary sensory, type IF, 615632
ATP13A2	100,0%	100,0%	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP1A1	100,0%	100,0%	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP7A	100,0%	100,0%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
B2M	100,0%	100,0%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B4GALNT1	100,0%	100,0%	Spastic paraplegia 26, autosomal recessive, 609195
BAG3	100,0%	100,0%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	100,0%	100,0%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BSCL2	100,0%	100,0%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
C19orf12	100,0%	100,0%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CADM3	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519
CCT5	100,0%	100,0%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
C1orf194	100,0%	100,0%	No OMIM Disease ID
CHCHD10	100,0%	100,0%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CNTNAP1	100,0%	100,0%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
COA3	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA7	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COL6A5	100,0%	100,0%	No OMIM Disease ID

COMP	100,0%	100,0%	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400
COX20	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX6A1	100,0%	100,0%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CTDP1	100,0%	100,0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CYP27A1	100,0%	100,0%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	100,0%	100,0%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	100,0%	100,0%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DARS2	100,0%	100,0%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF8	100,0%	100,0%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCTN1	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605
DCTN2	100,0%	100,0%	No OMIM Disease ID
DGAT2	100,0%	100,0%	No OMIM Disease ID
DHTKD1	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DNAH10	100,0%	100,0%	Spermatogenic failure 56, 619515
DNAJB2	100,0%	100,0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	100,0%	100,0%	No OMIM Disease ID
DNAJC3	100,0%	100,0%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	100,0%	100,0%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	100,0%	100,0%	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT1	100,0%	99,7%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DRP2	100,0%	100,0%	No OMIM Disease ID
DST	95,6%	95,6%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DYNC1H1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Intellectual developmental disorder, autosomal dominant 13, 614563

EGR2	100,0%	100,0%	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1D, 607678 Hypomyelinating neuropathy, congenital, 1, 605253
ELF2	100,0%	100,0%	No OMIM Disease ID
ELOVL5	100,0%	100,0%	Spinocerebellar ataxia 38, 615957
ELP1	100,0%	100,0%	Dysautonomia, familial, 223900 Medulloblastoma, 155255
EMILIN1	100,0%	100,0%	No OMIM Disease ID
ERBB2	100,0%	100,0%	Gastric cancer, somatic, 613659 Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 ?Visceral neuropathy, familial, 2, autosomal recessive, 619465 Glioblastoma, somatic, 137800
ERBB3	100,0%	100,0%	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
EXOSC8	100,0%	100,0%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100,0%	100,0%	Pontocerebellar hypoplasia, type 1D, 618065
FAM126A	100,0%	100,0%	Leukodystrophy, hypomyelinating, 5, 610532
FBLN5	91,8%	91,8%	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	100,0%	100,0%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FBXO38	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IID, 615575
FGD4	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	100,0%	100,0%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FLVCR1	100,0%	100,0%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FXN	100,0%	100,0%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GALC	100,0%	100,0%	Krabbe disease, 245200
GAN	100,0%	100,0%	Giant axonal neuropathy-1, 256850

GARS1	100,0%	100,0%	Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794 Charcot-Marie-Tooth disease, type 2D, 601472
GBA2	100,0%	100,0%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100,0%	100,0%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GBF1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GDAP1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, type 4A, 214400
GJB1	100,0%	100,0%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB3	100,0%	100,0%	Deafness, digenic, GJB2/GJB3, 220290 Deafness, autosomal dominant 2B, 612644 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, Deafness, autosomal dominant, with peripheral neuropathy,
GLA	91,3%	91,3%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLE1	100,0%	100,0%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GNB4	100,0%	100,0%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNE	100,0%	100,0%	Sialuria, 269921 Nonaka myopathy, 605820
GSN	100,0%	100,0%	Amyloidosis, Finnish type, 105120
HADHA	100,0%	100,0%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	100,0%	100,0%	Trifunctional protein deficiency, 609015
HARS1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HINT1	100,0%	100,0%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	100,0%	100,0%	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700

HMBS	100,0%	100,0%	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000
HOXD10	100,0%	100,0%	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPB1	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	100,0%	100,0%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
IFRD1	100,0%	100,0%	No OMIM Disease ID
IGHMBP2	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INF2	100,0%	100,0%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ITPR3	100,0%	100,0%	No OMIM Disease ID
JAG1	100,0%	100,0%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KARS1	100,0%	100,0%	Deafness, autosomal recessive 89, 613916 Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KBTBD13	100,0%	100,0%	Nemaline myopathy 6, autosomal dominant, 609273
KIF1A	98,0%	98,0%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	100,0%	100,0%	Pheochromocytoma, 171300 Charcot-Marie-Tooth disease, type 2A1, 118210
KIF5A	100,0%	100,0%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KLC2	100,0%	100,0%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
LAMA2	100,0%	100,0%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LITAF	100,0%	100,0%	Charcot-Marie-Tooth disease, type 1C, 601098

LMNA	100,0%	100,0%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Restrictive dermopathy 2, 619793 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LRIG3	100,0%	100,0%	No OMIM Disease ID
LRSAM1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MAG	100,0%	100,0%	Spastic paraplegia 75, autosomal recessive, 616680
MARS1	100,0%	100,0%	Interstitial lung and liver disease, 615486 ?Trichothiodystrophy 9, nonphotosensitive, 619692 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MCM3AP	100,0%	100,0%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MED25	100,0%	100,0%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MFN2	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MME	98,0%	98,0%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MORC2	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPV17	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	100,0%	100,0%	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
MTMR2	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4B1, 601382
C12orf65	100,0%	100,0%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559

MYH14	100,0%	100,0%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYO1A	100,0%	100,0%	No OMIM Disease ID
NAGLU	100,0%	100,0%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NARS1	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091
NDRG1	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4D, 601455
NDUFA9	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NEFH	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	100,0%	100,0%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NEMF	100,0%	100,0%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NFASC	100,0%	100,0%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NGF	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NIPA1	100,0%	100,0%	Spastic paraplegia 6, autosomal dominant, 600363
NMNAT2	100,0%	100,0%	No OMIM Disease ID
NTRK1	100,0%	100,0%	Insensitivity to pain, congenital, with anhidrosis, 256800
PDK3	100,0%	100,0%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDXK	100,0%	99,8%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100,0%	100,0%	Spinocerebellar ataxia 23, 610245
PEX1	100,0%	100,0%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100,0%	100,0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX16	100,0%	100,0%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX7	91,3%	91,3%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHYH	100,0%	100,0%	Refsum disease, 266500
PIEZO2	100,0%	100,0%	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700

PLA2G6	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLD3	100,0%	100,0%	?Spinocerebellar ataxia 46, 617770
PLEKHG5	96,3%	96,3%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PMM2	100,0%	100,0%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100,0%	100,0%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100,0%	100,0%	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
PNKP	100,0%	100,0%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPT1	100,0%	100,0%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	100,0%	100,0%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	100,0%	100,0%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POLR3B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
PRDM12	96,2%	94,0%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRNP	100,0%	100,0%	Spongiform encephalopathy with neuropsychiatric features, 606688 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400
PRPS1	100,0%	100,0%	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661

			Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PRX	97,9%	96,8%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100,0%	100,0%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
RAB7A	100,0%	100,0%	Charcot-Marie-Tooth disease, type 2B, 600882
REEP1	100,0%	100,0%	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
RETREG1	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RNF170	100,0%	100,0%	Ataxia, sensory, 1, autosomal dominant, 608984 Spastic paraplegia 85, autosomal recessive, 619686
SACS	100,0%	100,0%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	100,0%	100,0%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SBF1	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCARB2	100,0%	100,0%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN10A	100,0%	100,0%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100,0%	100,0%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	100,0%	100,0%	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
SCO2	100,0%	100,0%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	100,0%	100,0%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEPTIN9	100,0%	100,0%	Amyotrophy, hereditary neuralgic, 162100
SETX	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGPL1	100,0%	100,0%	Nephrotic syndrome, type 14, 617575

SH3TC2	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SIGMAR1	100,0%	100,0%	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373
SLC12A6	100,0%	100,0%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC25A19	100,0%	100,0%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A46	100,0%	100,0%	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC52A2	100,0%	100,0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100,0%	100,0%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A7	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC9A3R1	100,0%	100,0%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SORD	98,6%	96,1%	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912
SOX10	100,0%	100,0%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPAST	100,0%	100,0%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	100,0%	100,0%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG7	100,0%	100,0%	Spastic paraplegia 7, autosomal recessive, 607259
SPTAN1	100,0%	100,0%	Developmental and epileptic encephalopathy 5, 613477
SPTBN4	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	100,0%	100,0%	No OMIM Disease ID
SURF1	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SYT2	100,0%	100,0%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
TBCE	100,0%	100,0%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TDP1	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250

TDRKH	100,0%	100,0%	No OMIM Disease ID
TECPR2	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TFG	100,0%	100,0%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TRIM2	93,9%	93,9%	Charcot-Marie-Tooth disease, type 2R, 615490
TRPV4	100,0%	100,0%	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	94,6%	94,6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TUBB2A	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB3	100,0%	100,0%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	100,0%	100,0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TYMP	100,0%	100,0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBA5	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UCHL1	100,0%	100,0%	Spastic paraplegia 79, autosomal recessive, 615491
UQCRC1	100,0%	100,0%	Parkinsonism with polyneuropathy, 619279
VCP	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VRK1	100,0%	100,0%	Pontocerebellar hypoplasia type 1A, 607596
VWA1	100,0%	100,0%	Neuropathy, hereditary motor, with myopathic features, 619216
WARS1	100,0%	100,0%	Neuronopathy, distal hereditary motor, type IX, 617721
WNK1	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
XRCC1	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 26, 617633

YARS1	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 Charcot-Marie-Tooth disease, dominant intermediate C, 608323
ZFYVE26	100,0%	100,0%	Spastic paraplegia 15, autosomal recessive, 270700

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.
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.
TWIST 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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors