NEUROPATHIES GENE PANEL DG 3.1.0 (212 genes)

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Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS1	100	99,9	100	100	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
ABCA1	99,9	99,1	100	100	HDL deficiency, familial, 1, 604091 Tangier disease, 205400
ABCD1	75,8	71,6	100	100	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABHD12	91,2	85,2	100	99,4	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ADPRS	100	99,8	100	100	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	95	91,1	100	99,9	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGRN	96,9	92,6	100	99,9	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGTPBP1	96	94,1	100	100	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AIFM1	99,9	98,8	100	100	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
AMACR	100	100	100	100	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
APTX	94,9	92,4	100	100	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARHGEF10	99,8	98	100	100	?Slowed nerve conduction velocity, AD, 608236
ARHGEF28	99,2	94,4	100	100	No OMIM disease ID
ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ATAD3A	91,9	83,2	100	100	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
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

ATP13A2	100	99,5	100	100	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP7A	99	96,9	100	100	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
BAG3	100	100	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	100	99,7	100	100	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BSCL2	100	100	100	100	Neuropathy, distal hereditary motor, type VC, 619112 Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
C12orf65	99,8	98,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C19orf12	100	99,8	100	100	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1orf194	100	99,6	100	100	No OMIM disease ID
CADM3	100	99,9	100	100	No OMIM disease ID
CCT5	100	99,7	100	100	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHCHD10	59,1	43,9	100	100	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CNTNAP1	100	99,8	100	100	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
COA3	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA7	100	100	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COL6A5	99,9	99,5	100	100	No OMIM disease ID
COX20	97,8	88,3	100	100	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX6A1	100	99,5	100	100	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CTDP1	88,4	84,3	100	99,4	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CYP27A1	98,9	96,7	100	100	Cerebrotendinous xanthomatosis, 213700
DCAF8	100	99,9	100	100	?Giant axonal neuropathy 2, autosomal dominant, 610100

DCTN1	100	98,8	100	100	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605 Neuronopathy, distal hereditary motor, type VIIB, 607641
DCTN2	100	99,7	100	100	No OMIM disease ID
DGAT2	99,1	95,5	100	100	No OMIM disease ID
DHTKD1	99,9	98,9	100	100	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAH10	99,9	99,4	100	100	No OMIM disease ID
DNAJB2	100	100	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	95,7	91,3	100	100	No OMIM disease ID
DNAJC3	100	99,7	100	100	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	99,9	98,5	100	100	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	98,1	94,5	100	100	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DNMT1	99,2	99	99,7	99,2	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DRP2	99,1	96,5	100	99,9	No OMIM disease ID
DST	95,5	95	95,6	95,6	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
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
EGR2	100	100	100	100	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
ELOVL5	100	99,8	100	100	Spinocerebellar ataxia 38, 615957
ELP1	99,8	99	100	100	Dysautonomia, familial, 223900
EMILIN1	99,3	89,8	100	100	No OMIM disease ID
EXOSC8	97,9	91,2	100	100	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 1D, 618065
FAM126A	100	99,4	100	100	Leukodystrophy, hypomyelinating, 5, 610532
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
FBXO38	99,9	99,3	100	100	Neuronopathy, distal hereditary motor, type IID, 615575
FGD4	99,9	99,4	100	100	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	100	99,8	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FLVCR1	100	98,9	100	100	Ataxia, posterior column, with retinitis pigmentosa, 609033
FXN	95,5	80,1	100	100	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GALC	99,8	98,3	100	100	Krabbe disease, 245200
GAN	100	99,6	100	100	Giant axonal neuropathy-1, 256850
GARS1	99,9	99,1	100	100	Charcot-Marie-Tooth disease, type 2D, 601472 Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794
GBE1	100	99,6	100	100	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GBF1	98,3	98	100	100	No OMIM disease ID
GDAP1	99,8	99,3	100	100	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706
GJB1	100	100	100	100	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB3	100	100	100	100	Deafness, autosomal dominant 2B, 612644 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, 0 Deafness, autosomal dominant, with peripheral neuropathy, 0
GLA	91,1	88,2	91,3	91,3	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLE1	100	100	100	100	Congenital arthrogryposis with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GNB4	100	100	100	100	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNE	100	99,7	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GSN	95,8	93,5	99,9	99,3	Amyloidosis, Finnish type, 105120

HADHA	97,2	91,6	100	100	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016
HADHB	98,8	89,7	100	100	Trifunctional protein deficiency, 609015
HARS1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HINT1	98,3	89,3	100	100	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	100	100	100	100	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HMBS	99,9	99,4	100	100	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HOXD10	100	100	100	100	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPB1	98,8	91,6	100	100	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	100	100	100	100	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	100	100	100	100	Neuronopathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
IFRD1	99,7	98,6	100	100	No OMIM disease ID
IGHMBP2	98,8	95,1	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INF2	86,7	83,8	100	100	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ITPR3	100	99,7	100	100	{Diabetes, type 1, susceptibility to}, 222100
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KARS1	100	99,9	100	100	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KBTBD13	99,8	95,8	100	100	Nemaline myopathy 6, autosomal dominant, 609273

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
KIF1B	100	99,6	100	100	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700
KIF5A	100	99,9	100	100	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KLC2	99,2	97,9	100	100	Spastic paraplegia, optic atrophy, and neuropathy, 609541
LAMA2	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LITAF	98,2	92,7	100	100	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	97,4	91,9	100	100	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LRIG3	99,8	98,8	100	99,8	No OMIM disease ID
LRSAM1	100	99,9	100	100	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MAG	100	100	100	100	Spastic paraplegia 75, autosomal recessive, 616680
MARS1	99,7	97,4	100	100	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MCM3AP	99,9	99,1	100	100	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MED25	100	99,8	100	100	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MFN2	100	99,9	100	100	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MME	99,8	98,7	98	98	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018

MORC2	100	99,8	100	100	Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPV17	100	97,2	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
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
MTMR2	100	99	100	100	Charcot-Marie-Tooth disease, type 4B1, 601382
MYH14	98,4	94	100	100	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYO1A	100	99,8	100	100	No OMIM disease ID
NAGLU	92,9	89,9	99,9	99,2	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NARS1	100	100	100	100	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	100	100	100	Charcot-Marie-Tooth disease, type 4D, 601455
NDUFA9	99,9	96,5	100	100	Mitochondrial complex I deficiency, nuclear type 26, 618247
NEFH	93,4	84,5	100	100	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	99,9	98,2	100	100	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NEMF	100	99,2	100	100	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NFASC	100	99,9	100	100	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NGF	100	100	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NIPA1	100	100	99,8	98,5	Spastic paraplegia 6, autosomal dominant, 600363
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
PDK3	99,5	97,2	100	100	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDXK	79,3	76,6	99,4	96,7	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100	100	100	100	Spinocerebellar ataxia 23, 610245

PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,8	89,7	100	99,9	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX16	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	100	99,6	100	100	Refsum disease, 266500
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
PLA2G6	92,2	90,7	92,3	92,3	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLD3	99,9	99,1	100	100	?Spinocerebellar ataxia 46, 617770
PLEKHG5	95,3	91,1	96,3	96,2	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100	100	100	100	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
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
PNKP	100	100	100	100	Ataxia-oculomotor apraxia 4, 616267 ?Charcot-Marie-Tooth disease, type 2B2, 605589 Microcephaly, seizures, and developmental delay, 613402
PNPT1	97,7	89,7	100	100	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	100	99,3	100	100	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450

POLG2	99,6	98	100	99,9	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528
POLR3B	99,9	98,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
PRDM12	90,8	88	93,4	91,7	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRNP	100	100	100	100	Insomnia, fatal familial, 600072 {Kuru, susceptibility to}, 245300 Huntington disease-like 1, 603218 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Spongiform encephalopathy with neuropsychiatric features, 606688
PRPS1	86,4	86,4	100	100	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PRX	96	95,5	96,5	96,1	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100	100	100	100	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
RAB7A	100	99,9	100	100	Charcot-Marie-Tooth disease, type 2B, 600882
REEP1	78,7	76,1	100	100	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
RETREG1	98,8	95,1	100	100	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RNF170	99,6	97,6	100	100	Ataxia, sensory, 1, autosomal dominant, 608984
SACS	100	99,9	100	100	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	100	100	100	100	Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Ataxia-pancytopenia syndrome, 159550
SBF1	99	97,7	100	100	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	99,9	99,4	100	100	Charcot-Marie-Tooth disease, type 4B2, 604563
SCARB2	100	99,8	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN10A	100	99,6	100	100	Episodic pain syndrome, familial, 2, 615551
SCN11A	99,8	98,3	100	100	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548

SCN9A	99,3	97,9	100	100	Neuropathy, hereditary sensory and autonomic, type IID, 243000 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020
SCO2	100	100	100	100	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	100	99,2	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEPTIN9	100	99,9	100	100	Amyotrophy, hereditary neuralgic, 162100
SETX	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SH3TC2	100	99,7	100	100	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SIGMAR1	100	100	100	100	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SLC12A6	100	100	100	100	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC25A19	100	98,5	100	100	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A46	99,7	97,3	100	100	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC5A7	100	99,9	100	100	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SORD	90,3	89,1	97	93,6	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912
SOX10	99,9	97,9	100	100	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SPG11	100	99,3	100	100	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPTAN1	99,1	98,6	100	100	Developmental and epileptic encephalopathy 5, 613477
SPTBN4	97,3	91	100	100	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
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

SPTLC3	100	99,9	100	100	No OMIM disease ID
SURF1	89,4	88,2	100	100	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SYT2	99,9	99	100	100	Myasthenic syndrome, congenital, 7, presynaptic, 616040
TBCE	99,8	97,5	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TDP1	99,9	99,5	100	100	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDRKH	94,7	94,7	100	100	No OMIM disease ID
TFG	96,9	96,3	100	100	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TRIM2	93,9	93,3	93,9	93,9	Charcot-Marie-Tooth disease, type 2R, 615490
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
TUBB2A	97	95,7	100	100	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB3	98,3	96,9	100	100	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	100	100	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
UBA5	97,8	86,8	100	100	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UCHL1	99,8	92,5	100	100	Spastic paraplegia 79, autosomal recessive, 615491 {?Parkinson disease 5, susceptibility to}, 613643
UQCRC1	99,8	98,4	100	100	No OMIM disease ID

VCP	100	99,2	100	100	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954
VRK1	99,7	98,5	100	100	Pontocerebellar hypoplasia type 1A, 607596
VWA1	84,1	76,3	99,2	95,1	Neuropathy, hereditary motor, with myopathic features, 619216
WARS1	99,8	98,3	100	100	Neuronopathy, distal hereditary motor, type IX, 617721
WNK1	99,9	99,6	100	100	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
XRCC1	100	98,8	100	100	?Spinocerebellar ataxia, autosomal recessive 26, 617633
YARS1	100	99,9	100	100	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
ZFYVE26	100	99,1	100	100	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.
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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors