

NEUROPATHIES GENE PANEL DG 3.2.0 (228 genes)

Releasedate: 16-09-2021

<i>Gene</i>	<i>Agilent V5 covered >10x</i>	<i>Agilent V5 covered >20x</i>	<i>TWIST covered >10x</i>	<i>TWIST covered >20x</i>	<i>Associated Phenotype Description and OMIM disease ID</i>
AAAS	100	99,4	100	100	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100	99,7	100	99,9	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
ABCA1	99,8	97,8	100	100	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCD1	76	72,6	100	100	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	91,7	86	100	99,8	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ADPRS	100	99,9	100	100	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	94,6	86,3	100	100	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGRN	97,6	92,6	100	99,9	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGTPBP1	96,3	94,2	100	100	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AHNAK2	97,3	96,9	96,7	96,5	No OMIM disease ID
AIFM1	99,9	97,8	100	100	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
AMACR	100	100	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
APTX	94,1	90,6	100	100	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARHGEF10	99,6	97	100	100	?Slowed nerve conduction velocity, AD, 608236
ARHGEF28	99,3	95,3	100	100	No OMIM disease ID
ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ATAD3A	91,4	86,7	100	100	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810

ATL1	99,9	99,5	100	99,8	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	99,6	97,6	100	99,9	Neuropathy, hereditary sensory, type IF, 615632
ATP13A2	100	99,6	100	100	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP1A1	100	99,8	100	100	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP7A	98,7	96	100	99,9	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
B2M	100	100	100	100	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B4GALNT1	98,3	93,5	100	100	Spastic paraplegia 26, autosomal recessive, 609195
BAG3	100	99,7	100	100	Cardiomyopathy, dilated, 1HYPOGONADOTROPIC HYPOGONADISM, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	99,9	99,1	100	100	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BSCL2	100	99,9	100	100	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	99,8	100	100	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CADM3	100	99,7	100	100	No OMIM disease ID
CCT5	99,9	99,3	100	100	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
C1orf194	100	98,4	100	100	No OMIM disease ID
CHCHD10	57,8	42	100	100	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
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	99,2	100	100	No OMIM disease ID

COMP	93,8	92,4	100	100	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400
COX20	95,7	82,4	100	99,9	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX6A1	100	99,9	100	100	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CTDP1	88,7	85	100	99,8	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CYP27A1	99,7	98,1	100	100	Cerebrotendinous xanthomatosis, 213700
CYP2U1	95,3	92	100	99,9	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	98,1	92,7	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DARS2	94,8	93,8	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF8	100	99,4	100	100	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCTN1	99,8	98,4	100	100	Neuronopathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605
DCTN2	99,9	97,9	100	100	No OMIM disease ID
DGAT2	98,5	94,7	100	100	No OMIM disease ID
DHTKD1	99,8	98,8	100	100	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DNAH10	99,9	99,1	100	100	Spermatogenic failure 56, 619515
DNAJB2	100	100	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	95,5	89,4	100	100	No OMIM disease ID
DNAJC3	99,7	99,7	100	99,9	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	99,6	98,3	100	100	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	98,6	93,9	100	100	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	99,2	98,8	99,9	99,4	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DRP2	98,3	93,6	100	100	No OMIM disease ID
DST	95,3	94,5	95,6	95,6	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653

DYNC1H1	99,9	99,3	100	100	Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563
EGR2	100	100	100	100	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1D, 607678 Hypomyelinating neuropathy, congenital, 1, 605253
ELF2	99	94,8	100	100	No OMIM disease ID
ELOVL5	100	99,4	100	100	Spinocerebellar ataxia 38, 615957
ELP1	99,8	98,9	100	100	Dysautonomia, familial, 223900
EMILIN1	98,3	90,3	100	100	No OMIM disease ID
ERBB2	98,5	97,2	100	100	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	99,3	100	100	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
EXOSC8	98,7	90	100	100	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	99,3	94,7	100	99,9	Pontocerebellar hypoplasia, type 1D, 618065
FAM126A	99,5	99,4	100	100	Leukodystrophy, hypomyelinating, 5, 610532
FBLN5	91,8	91,7	91,8	91,8	Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 ?Cutis laxa, autosomal dominant 2, 614434
FBXO38	99,8	98,9	100	100	Neuronopathy, distal hereditary motor, type IID, 615575
FGD4	99,8	98,8	100	100	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	99,8	99,7	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FLVCR1	99,7	98,3	100	99,9	Ataxia, posterior column, with retinitis pigmentosa, 609033
FXN	98,3	84,7	100	100	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GALC	99,7	97,6	100	100	Krabbe disease, 245200

GAN	99,9	98,8	100	100	Giant axonal neuropathy-1, 256850
GARS1	99,9	99,4	100	100	Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794 Charcot-Marie-Tooth disease, type 2D, 601472
GBA2	100	99,5	100	100	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	99,9	99,7	100	100	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GBF1	98,3	97,7	100	100	No OMIM disease ID
GDAP1	99,7	98,4	100	100	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	100	100	100	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB3	100	100	100	100	Deafness, digenic, GJB2/GJB3, 220290 Deafness, autosomal dominant 2B, 612644 Erythrokeratoderma variabilis et progressiva 1, 133200 Deafness, autosomal recessive, Deafness, autosomal dominant, with peripheral neuropathy,
GLA	91	85,9	91,3	91,3	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLE1	100	99,9	100	100	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GNB4	99,8	99,6	100	100	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNE	100	99,5	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GSN	95,7	93,5	100	99,7	Amyloidosis, Finnish type, 105120
HADHA	95,5	88,3	100	100	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	97,7	87	100	99,9	Trifunctional protein deficiency, 609015
HARS1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HINT1	95,2	82,5	100	100	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200

HK1	100	99,9	100	100	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	98,4	100	100	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000
HOXD10	100	99,6	100	100	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950
HSD17B4	95,3	92,8	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPB1	99,1	92,1	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	99,9	99,2	100	100	No OMIM disease ID
IGHMBP2	99,3	96,9	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INF2	87,2	84,8	100	100	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ITPR3	100	99,6	100	100	No OMIM disease ID
JAG1	97,8	96,7	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KARS1	99,9	98,9	100	100	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	99,9	96,7	100	100	Nemaline myopathy 6, autosomal dominant, 609273
KIF1A	97,4	95,3	98	98	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	99,9	99,2	100	100	Pheochromocytoma, 171300 Charcot-Marie-Tooth disease, type 2A1, 118210
KIF5A	100	99,8	100	100	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187

KLC2	99,2	98	100	100	Spastic paraplegia, optic atrophy, and neuropathy, 609541
LAMA2	99,9	99,1	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LITAF	97,1	91,9	100	100	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	96,1	90,6	100	100	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Restrictive dermopathy, lethal, 275210 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 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	99,4	100	99,7	No OMIM disease ID
LRSAM1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MAG	100	100	100	100	Spastic paraplegia 75, autosomal recessive, 616680
MARS1	99	96,1	100	100	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MCM3AP	99,9	99,1	100	100	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MED25	100	99,9	100	99,9	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MFN2	100	99,8	100	100	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MME	99,7	98,6	98	97,9	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MORC2	100	99,5	100	100	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPV17	100	98,7	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	85,6	81,9	100	100	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	99,5	98,4	100	100	Charcot-Marie-Tooth disease, type 4B1, 601382
C12orf65	99	94,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MYH14	98,2	93,3	100	100	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYO1A	99,9	99,2	100	100	No OMIM disease ID
NAGLU	93,8	91,7	99,9	98,7	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
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	99,9	100	100	Charcot-Marie-Tooth disease, type 4D, 601455
NDUFA9	99,3	95,2	100	100	Mitochondrial complex I deficiency, nuclear type 26, 618247
NEFH	96,3	88,2	100	100	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	99,4	96,8	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	99,6	98,3	100	99,9	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NFASC	100	99,5	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	99,7	98,1	Spastic paraplegia 6, autosomal dominant, 600363
NMNAT2	100	98,8	100	100	No OMIM disease ID
NTRK1	99,9	98,5	100	100	Insensitivity to pain, congenital, with anhidrosis, 256800
PDK3	98,8	95,5	100	99,8	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDXK	79,4	77,1	99,9	99,1	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100	100	100	100	Spinocerebellar ataxia 23, 610245
PEX1	99,8	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100

PEX10	98,8	90,6	100	100	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX16	97,1	93,9	100	100	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX7	88	81	91,3	91,2	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHYH	100	98,9	100	100	Refsum disease, 266500
PIEZO2	99,8	99,2	100	100	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PLA2G6	92,1	90,7	92,3	92,3	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLD3	100	99,2	100	100	?Spinocerebellar ataxia 46, 617770
PLEKHG5	96,1	93,4	96,3	96,2	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PMM2	99,8	99,8	100	100	Congenital disorder of glycosylation, type Ia, 212065
PMP2	99,7	99,7	100	100	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100	100	100	100	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	100	100	100	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPT1	96,9	86,1	100	99,9	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	99,9	98,8	100	100	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	99,3	97,2	100	99,8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neurophthalmic type), 619425
POLR3B	99,7	97,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
PRDM12	91,7	89,6	92,8	91	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRNP	100	99,6	100	100	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	86,4	86,3	100	99,7	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	96,1	95,8	97,5	96,4	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100	99,6	100	100	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
RAB7A	100	100	100	100	Charcot-Marie-Tooth disease, type 2B, 600882
REEP1	78,6	76,4	100	100	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
RETREG1	99,1	96,1	100	100	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RNF170	98,2	94,2	100	100	Ataxia, sensory, 1, autosomal dominant, 608984
SACS	99,9	99,9	100	100	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	100	99,9	100	100	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SBF1	99,1	98	100	100	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	99,7	98,6	100	100	Charcot-Marie-Tooth disease, type 4B2, 604563
SCARB2	99,9	99,4	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
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 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	99,1	97	100	100	Erythralgia, 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	100	100	100	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	99,9	97,9	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100	99,9	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEPTIN9	100	99,5	100	100	Amyotrophy, hereditary neuralgic, 162100
SETX	99,8	99,6	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	99,4	100	100	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SIGMAR1	100	100	100	100	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373
SLC12A6	100	100	100	100	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC25A19	99,9	98	100	100	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A46	99,7	98,6	100	100	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A7	100	100	100	100	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC9A3R1	99,9	98,2	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SORD	90,6	89,4	98,4	95,1	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912
SOX10	99,9	97,2	100	100	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPAST	99,4	98,1	100	100	Spastic paraplegia 4, autosomal dominant, 182601

SPG11	99,8	99	100	100	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG7	90,4	86,7	100	100	Spastic paraplegia 7, autosomal recessive, 607259
SPTAN1	99,1	97,9	100	100	Developmental and epileptic encephalopathy 5, 613477
SPTBN4	98,1	92,1	100	100	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
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
SPTLC3	100	99,8	100	100	No OMIM disease ID
SURF1	89,5	88,1	100	100	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SYT2	100	98,8	100	100	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
TBCE	99,7	96,6	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TDP1	99,9	99,4	100	100	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDRKH	94,7	94,6	100	100	No OMIM disease ID
TECPR2	100	100	100	100	Spastic paraplegia 49, autosomal recessive, 615031
TFG	97	96	100	99,9	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TRIM2	93,8	93,5	93,9	93,9	Charcot-Marie-Tooth disease, type 2R, 615490
TRPV4	100	99,9	100	100	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	94,6	94,6	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430

TUBB2A	96,9	95,7	100	100	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB3	98,5	96,8	100	100	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TWINK	100	99,9	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
TYMP	100	99,4	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBA5	97,4	86,6	100	100	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UCHL1	99,3	90,5	100	100	Spastic paraplegia 79, autosomal recessive, 615491
UQCRC1	99,8	98,3	100	100	Parkinsonism with polyneuropathy, 619279
VCP	100	99,1	100	100	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	99,4	97,8	100	100	Pontocerebellar hypoplasia type 1A, 607596
VWA1	85,4	77,7	99,6	96,1	Neuropathy, hereditary motor, with myopathic features, 619216
WARS1	99,6	97,4	100	100	Neuronopathy, distal hereditary motor, type IX, 617721
WNK1	99,8	99,3	100	100	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
XRCC1	99,6	97,2	100	100	?Spinocerebellar ataxia, autosomal recessive 26, 617633
YARS1	100	99,8	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 Charcot-Marie-Tooth disease, dominant intermediate C, 608323
ZFYVE26	99,7	97,8	100	100	Spastic paraplegia 15, autosomal recessive, 270700

Gene symbols used follow HGNC 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 between {} signify risk factors
