NEUROPATHIES GENE PANEL DG 2.18 (172 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AARS	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
ABCA1	99,90%	99,10%	100%	100%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCD1	75,80%	71,60%	100%	100%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABHD12	98,70%	92,30%	100%	99,30%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ADPRHL2	100%	99,80%	100%	100%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AGRN	96,90%	92,60%	100%	99,90%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGTPBP1	96,00%	94,10%	100%	100%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AIFM1	99,90%	98,80%	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	94,90%	92,50%	100%	100%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARHGEF10	99,80%	98,00%	100%	100%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF28	99,20%	94,40%	100%	100%	No OMIM disease ID
ARSA	100%	99,80%	100%	100%	Metachromatic leukodystrophy, 250100
ATAD3A	91,90%	83,20%	100%	100%	Harel-Yoon syndrome, 617183 ?Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATL1	100%	99,70%	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	99,80%	98,30%	100%	100%	Neuropathy, hereditary sensory, type IF, 615632
ATP1A1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP7A	99,70%	97,50%	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%	99,70%	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%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
C12orf65	99,80%	98,50%	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C1orf194	100%	99,60%	100%	100%	No OMIM disease ID
CCT5	100%	99,70%	100%	100%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHCHD10	59,10%	43,90%	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%	99,80%	100%	100%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
COA7	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COX20	97,80%	88,30%	100%	100%	Mitochondrial complex IV deficiency, 220110
COX6A1	100%	99,50%	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CTDP1	88,40%	84,30%	100%	99,40%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CYP27A1	98,90%	96,70%	100%	100%	Cerebrotendinous xanthomatosis, 213700
DCAF8	100%	99,90%	100%	100%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCTN1	100%	98,80%	100%	100%	Perry syndrome, 168605 Neuronopathy, distal hereditary motor, type VIIB, 607641
DCTN2	100%	99,70%	100%	100%	No OMIM disease ID
DGAT2	99,10%	95,50%	100%	100%	No OMIM disease ID
DHTKD1	99,90%	98,90%	100%	100%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAH10	99,90%	99,40%	100%	100%	No OMIM disease ID
DNAJB2	100%	100%	100%	100%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	95,70%	91,30%	100%	100%	No OMIM disease ID
DNM2	98,10%	94,50%	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	99,20%	99,00%	99,70%	99,20%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121

DRP2	99,10%	96,50%	100%	99,90%	No OMIM disease ID
DST	99,90%	99,40%	100%	100%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DYNC1H1	99,90%	99,40%	100%	100%	Mental retardation, autosomal dominant 13, 614563 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
EGR2	100%	100%	100%	100%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
ELOVL5	100%	99,80%	100%	100%	Spinocerebellar ataxia 38, 615957
ELP1	99,80%	99,00%	100%	100%	Dysautonomia, familial, 223900
EXOSC8	97,90%	91,20%	100%	100%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 1D, 618065
FAM126A	100%	99,40%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
FBLN5	91,80%	91,80%	91,80%	91,80%	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	99,90%	99,30%	100%	100%	Neuronopathy, distal hereditary motor, type IID, 615575
FGD4	99,90%	99,40%	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	100%	99,80%	100%	100%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FLVCR1	100%	98,90%	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
GALC	99,80%	98,30%	100%	100%	Krabbe disease, 245200
GAN	100%	99,60%	100%	100%	Giant axonal neuropathy-1, 256850
GARS	99,90%	99,10%	100%	100%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuronopathy, distal hereditary motor, type VA, 600794
GBE1	100%	99,60%	100%	100%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GDAP1	99,80%	99,30%	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	99,80%	96,60%	100%	100%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLE1	100%	100%	100%	100%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GNB4	100%	100%	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GSN	95,80%	93,50%	99,90%	99,30%	Amyloidosis, Finnish type, 105120
HADHA	97,10%	91,30%	100%	100%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HADHB	98,80%	89,70%	100%	100%	Trifunctional protein deficiency, 609015
HARS	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HINT1	98,30%	89,30%	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	99,90%	99,40%	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	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPB1	98,80%	91,60%	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	99,70%	98,60%	100%	100%	No OMIM disease ID
IGHMBP2	98,80%	95,10%	100%	100%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INF2	86,70%	83,80%	100%	100%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455

KARS	100%	99,90%	100%	100%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KBTBD13	99,80%	95,80%	100%	100%	Nemaline myopathy 6, autosomal dominant, 609273
KIF1A	99,40%	97,10%	100%	100%	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	100%	99,60%	100%	100%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210
KIF5A	100%	99,90%	100%	100%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
LITAF	98,20%	92,70%	100%	100%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	97,40%	91,90%	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	99,80%	98,80%	100%	99,80%	No OMIM disease ID
LRSAM1	100%	99,90%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MARS	99,70%	97,40%	100%	100%	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MCM3AP	99,90%	99,10%	100%	100%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MED25	100%	99,80%	100%	100%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MFN2	100%	99,90%	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	99,80%	98,70%	98,00%	98,00%	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MORC2	100%	99,80%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPV17	100%	97,20%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810

MPZ	100%	100%	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
MTMR 2	100%	99,00%	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MYH14	98,40%	94,00%	100%	100%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYO1A	100%	99,80%	100%	100%	No OMIM disease ID
NAGLU	92,90%	89,90%	99,90%	99,20%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NDRG1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, type 4D, 601455
NDUFA9	99,90%	96,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NEFH	93,40%	84,50%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	99,90%	98,20%	100%	100%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NGF	100%	100%	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NIPA1	100%	100%	99,80%	98,50%	Spastic paraplegia 6, autosomal dominant, 600363
NMNAT2	99,90%	98,90%	100%	100%	No OMIM disease ID
NTRK1	99,80%	98,20%	100%	100%	Insensitivity to pain, congenital, with anhidrosis, 256800
PDK3	99,50%	97,20%	100%	100%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDXK	79,30%	76,60%	99,40%	96,70%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100%	100%	100%	100%	Spinocerebellar ataxia 23, 610245
PEX1	99,90%	99,40%	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX7	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	100%	99,60%	100%	100%	Refsum disease, 266500
PIEZO2	100%	99,50%	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	99,90%	98,30%	100%	100%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLD3	99,90%	99,10%	100%	100%	?Spinocerebellar ataxia 46, 617770
PLEKHG5	98,90%	94,60%	100%	99,90%	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	99,20%	95,20%	95,40%	88,50%	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 Microcephaly, seizures, and developmental delay, 613402
PNPT1	97,70%	89,70%	100%	100%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	100%	99,30%	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
PRDM12	90,80%	88,00%	93,40%	91,70%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRPS1	100%	100%	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	100%	100%	100%	100%	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%	99,90%	100%	100%	Charcot-Marie-Tooth disease, type 2B, 600882
REEP1	78,70%	76,10%	100%	100%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751

RETREG1	98,80%	95,10%	100%	100%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RNF170	99,60%	97,60%	100%	100%	Ataxia, sensory, 1, autosomal dominant, 608984
SACS	100%	100%	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SBF1	99,00%	97,70%	100%	100%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	99,90%	99,40%	100%	100%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN10A	100%	99,70%	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN11A	99,80%	98,30%	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	99,30%	97,80%	100%	100%	Small fiber neuropathy, 133020 HSAN2D, autosomal recessive, 243000 Paroxysmal extreme pain disorder, 167400 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SCO2	100%	100%	100%	100%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCYL1	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEPT9	100%	99,90%	100%	100%	Amyotrophy, hereditary neuralgic, 162100
SETX	100%	99,80%	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%	99,70%	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
SLC25A46	99,70%	97,30%	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%	99,90%	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SOX10	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266

SPG11	100%	99,30%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPTBN4	97,30%	91,00%	100%	100%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	99,20%	95,40%	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%	99,90%	100%	100%	No OMIM disease ID
SURF1	89,40%	88,20%	100%	100%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
SYT2	99,90%	99,00%	100%	100%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
TBCE	99,80%	97,30%	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TDP1	99,90%	99,50%	100%	100%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TFG	96,90%	96,30%	100%	100%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TRIM2	93,90%	93,30%	93,90%	93,90%	Charcot-Marie-Tooth disease, type 2R, 615490
TRPV4	100%	99,90%	100%	100%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 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	94,60%	94,60%	94,60%	94,60%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TUBB3	98,30%	96,90%	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
VCP	100%	99,20%	100%	100%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VRK1	99,70%	98,50%	100%	100%	Pontocerebellar hypoplasia type 1A, 607596

WARS	99,80%	98,30%	100%	100%	Neuronopathy, distal hereditary motor, type IX, 617721
WNK1	99,90%	99,60%	100%	100%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
YARS	100%	99,90%	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.
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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors