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POLYNEUROPATHIES PANEL[1] DG-5.1.0 (248 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
AAAS	100%	100%	100%	100%	99.7%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100%	100%	100%	100%	99.4%	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%	100%	100%	100%	99.8%	Tangier disease, 205400;HDL deficiency, familial, 1, 604091
ABCD1	89.3%	86.3%	98.7%	89.2%	70.9%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABHD12	100%	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACOX1	97.9%	97.9%	100%	100%	99.7%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ADPRS	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170

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AFG3L2	94.2%	92.2%	100%	100%	99.5%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AGRN	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGTPBP1	96.5%	96.5%	100%	100%	99.7%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AHNAK2	97.8%	97.7%	95.6%	94.1%	91.1%	
AIFM1	100%	100%	99.3%	90.3%	72.6%	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%	99.8%	99.4%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
APTX	100%	100%	100%	100%	99.7%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARHGAP19	94.5%	94.1%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2KK, 621466
ARHGEF10	100%	100%	100%	100%	99.7%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF28	100%	100%	100%	100%	99.7%	

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ARSA	100%	100%	100%	99.9%	99%	Metachromatic leukodystrophy, 250100
ATAD3A	100%	100%	100%	99.9%	98.7%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATL1	100%	100%	100%	100%	99.7%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATL3	100%	100%	100%	100%	99.7%	Neuropathy, hereditary sensory, type IF, 615632
ATP13A2	100%	100%	100%	100%	99.5%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP1A1	97.2%	95.4%	100%	100%	99.6%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP7A	95%	94.8%	99.4%	92.2%	74.7%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
B2M	100%	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600
B4GALNT1	100%	100%	100%	100%	99.5%	Spastic paraplegia 26, autosomal recessive, 609195

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BAG3	100%	100%	100%	100%	99.5%	?Neuronopathy, distal hereditary motor, autosomal dominant 15, 621094;Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954;Charcot-Marie-Tooth disease, axonal, type 2JJ, 621095
BICD2	100%	100%	100%	100%	99.4%	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%	99.6%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
C19orf12	100%	100%	100%	99.9%	98.7%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
CADM3	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519
CCT5	100%	100%	100%	100%	99.5%	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840

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CD59	89.3%	89.3%	100%	100%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CFAP276	100%	100%	100%	100%	99.7%	
CHCHD10	100%	100%	100%	100%	99.5%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CNTNAP1	100%	100%	100%	100%	99.1%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186
COA3	100%	100%	100%	100%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA7	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COL6A5	100%	100%	100%	100%	99.8%	
COMP	96.8%	96.3%	100%	99.9%	99.3%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
COX18	100%	100%	100%	99.9%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 25, 621487;Charcot-Marie-Tooth disease, axonal, type 2MM, 621488
COX20	100%	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 11, 619054

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COX6A1	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CRYAB	100%	100%	100%	100%	99.7%	Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
CTDP1	90.8%	90.5%	100%	100%	99.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CYP27A1	100%	100%	100%	100%	99.5%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	100%	100%	100%	99.9%	98.8%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	100%	100%	100%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812
DARS2	100%	100%	100%	100%	99.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485
DCAF8	96%	96%	100%	100%	99.7%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCTN1	100%	100%	100%	100%	99%	Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641
DCTN2	100%	100%	100%	100%	99.5%	

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DGAT2	100%	100%	100%	100%	99.5%	
DHTKD1	92.7%	92.7%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DNAJB2	100%	100%	100%	99.9%	99.5%	Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881
DNAJB5	100%	100%	100%	100%	99.3%	
DNAJC3	92.1%	92.1%	100%	100%	99.8%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	93.3%	91.7%	100%	99.9%	99.4%	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%	99.3%	100%	100%	99.4%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DRP2	100%	100%	98.8%	88.9%	69.3%	

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DST	100%	100%	100%	100%	99.7%	Congenital myopathy 29 with contractures, 621510;Neuropathy, hereditary sensory and autonomic, type VI, 614653;Lethal congenital contracture syndrome 12, 621511;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DYNC1H1	99.3%	99.3%	100%	99.9%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563
EGR2	100%	100%	100%	100%	99.6%	Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253
ELF2	100%	100%	100%	100%	99.7%	
ELOVL5	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia 38, 615957
ELP1	100%	100%	100%	100%	99.8%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900
EMILIN1	100%	99.7%	100%	100%	99.3%	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908

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ERBB2	100%	100%	100%	100%	99.4%	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%	100%	100%	99.9%	98.9%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ETFDH	94.1%	92.3%	100%	100%	99.9%	Glutaric acidemia IIC, 231680
EXOSC8	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100%	100%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 1D, 618065
FAT3	100%	100%	100%	100%	99.8%	
FBLN5	92.8%	92.8%	100%	99.9%	99.4%	Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434
FBN2	99.2%	99.2%	100%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

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FBXO38	100%	100%	100%	100%	99.9%	Neuronopathy, distal hereditary motor, autosomal dominant 6, 615575
FGD4	100%	100%	100%	100%	99.8%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	98.4%	98.4%	100%	100%	99.9%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060
FXN	100%	100%	100%	100%	98.9%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
GALC	100%	100%	100%	100%	99.6%	Krabbe disease, 245200
GAN	100%	100%	100%	99.9%	99.5%	Giant axonal neuropathy-1, 256850
GARS1	98.9%	98.9%	100%	100%	99.8%	Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472
GBA2	100%	100%	100%	99.9%	99.4%	Spastic paraplegia 46, autosomal recessive, 614409

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GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GBF1	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GDAP1	86.2%	86.2%	98.3%	98%	97.1%	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%	97.9%	84.9%	64.7%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB3	100%	100%	100%	100%	99.5%	Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644
GLA	91.4%	91.4%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLE1	100%	100%	100%	100%	99.7%	Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890
GNB4	100%	100%	100%	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185

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GNE	100%	100%	100%	100%	99.8%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820
GSN	92.1%	90%	100%	99.8%	98.9%	Amyloidosis, Finnish type, 105120
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HARS1	93.2%	93.2%	100%	99.9%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504
HINT1	100%	100%	100%	100%	99.8%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	98.2%	96.5%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelopmental disorder with visual defects and brain anomalies, 618547

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HMBS	100%	100%	100%	100%	99.9%	Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000
HNRNPA1	100%	100%	100%	100%	99.7%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424;?Myopathy, distal, 3, 610099;Amyotrophic lateral sclerosis 20, 615426
HOXD10	100%	100%	100%	100%	98.6%	Vertical talus, congenital, 192950;Charcot-Marie-Tooth disease, foot deformity of, 192950
HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSPB1	100%	100%	100%	100%	99.1%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634
HSPB3	100%	100%	100%	100%	99.2%	?Neuronopathy, distal hereditary motor, autosomal dominant 4, 613376

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HSPB8	100%	100%	100%	100%	99.7%	Neuronopathy, distal hereditary motor, autosomal dominant 2, 158590;Charcot-Marie-Tooth disease, axonal, type 2L, 608673;Myopathy, myofibrillar, 13, with rimmed vacuoles, 621078
HYCC1	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
IARS2	100%	99.7%	100%	99.9%	99.6%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IFRD1	100%	100%	100%	100%	99.6%	
IGHMBP2	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320
INF2	100%	100%	99.9%	99.5%	98.1%	Glomerulosclerosis, focal segmental, 5, 613237;Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INSC	94.4%	94.4%	100%	100%	99.5%	
ITPR3	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111;Immunodeficiency 133 with ectodermal dysplasia with or without peripheral neuropathy, 621254

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JAG1	100%	100%	100%	100%	99.6%	?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	94.3%	94.3%	100%	100%	99.8%	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%	100%	100%	100%	99.1%	Nemaline myopathy 6, autosomal dominant, 609273
KIF1A	100%	100%	100%	100%	99.3%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
KIF1B	94.9%	94.9%	100%	100%	99.6%	{Neuroblastoma, susceptibility to, 1}, 256700;Charcot-Marie-Tooth disease, type 2A1, 118210

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KIF5A	100%	100%	100%	100%	99.5%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187
KLC2	100%	100%	100%	100%	99.4%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
LAMA2	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LITAF	100%	100%	100%	100%	99.3%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	100%	100%	100%	99.9%	99.3%	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

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LRSAM1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MAG	100%	100%	100%	100%	99.3%	Spastic paraplegia 75, autosomal recessive, 616680
MARS1	89.3%	88.7%	100%	100%	99.5%	Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MCM3AP	100%	100%	100%	100%	99.5%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MFN2	100%	100%	100%	100%	99.7%	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087;Hereditary motor and sensory neuropathy VIA, 601152
MME	97.7%	97.7%	100%	100%	99.7%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MORC2	100%	100%	100%	100%	99.3%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090

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MPV17	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	100%	100%	100%	100%	98.9%	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%	100%	100%	99.9%	99.6%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTRFR	100%	100%	100%	100%	99.7%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MYH14	100%	100%	100%	99.9%	98.8%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652
MYO9B	100%	100%	100%	100%	99.2%	
MYOT	100%	100%	100%	100%	99.8%	Myopathy, myofibrillar, 3, 609200

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NAGLU	100%	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NARS1	100%	100%	100%	100%	99.5%	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%	99.7%	Charcot-Marie-Tooth disease, type 4D, 601455
NDUFA9	87%	85.2%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFS6	94.7%	86.1%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NEFH	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924;{?Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	100%	100%	100%	99.9%	98.8%	Charcot-Marie-Tooth disease, type 1F, 607734;Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684

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NEMF	100%	100%	100%	99.8%	99.3%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NFASC	100%	100%	100%	99.9%	99.5%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NGF	100%	100%	100%	100%	98.9%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NHERF1	85.8%	85.8%	100%	100%	99.2%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
NIPA1	100%	100%	99.8%	99.2%	97.4%	Spastic paraplegia 6, autosomal dominant, 600363
NMNAT2	100%	100%	100%	100%	99.7%	
NRG1	100%	100%	100%	99.9%	99.3%	
NTRK1	100%	100%	100%	99.9%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800
PDHA1	91.4%	89.5%	99.3%	91.4%	72.9%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDK3	100%	100%	99.1%	91%	73.3%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDXK	91.6%	88.3%	100%	100%	99.3%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia 23, 610245

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PEX1	100%	100%	100%	100%	99.9%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	100%	99.6%	98.7%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX16	100%	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500
PIEZO2	100%	100%	100%	100%	99.7%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PIGB	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 80, 618580
PIGG	100%	100%	100%	100%	99.5%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917

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PLA2G6	100%	100%	100%	100%	99.4%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLD3	100%	100%	100%	100%	99.3%	?Spinocerebellar ataxia 46, 617770
PLEKHG5	100%	100%	100%	99.9%	99.3%	Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067;Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100%	100%	100%	100%	99.7%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100%	100%	100%	100%	99.7%	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%	99.9%	99.2%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402

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PNPT1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POLG	100%	100%	100%	100%	99.8%	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	95.8%	95.8%	100%	100%	99.5%	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

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POLR3A	100%	100%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
PRDM12	97.1%	92.6%	100%	99.9%	98.8%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRNP	100%	100%	100%	100%	99.8%	Spongiform encephalopathy with neuropsychiatric features, 606688;Gerstmann-Straussler disease, 137440;Huntington disease-like 1, 603218;Insomnia, fatal familial, 600072;{Kuru, susceptibility to}, 245300;Cerebral amyloid angiopathy, PRNP-related, 137440;Creutzfeldt-Jakob disease, 123400
PRPS1	100%	100%	99%	89.8%	71.8%	Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661

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PRX	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 4F, 614895;Dejerine-Sottas disease, 145900
PSAP	99.6%	97.1%	100%	100%	99.4%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491
RAB7A	89.2%	89.2%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 2B, 600882
RCC1	100%	100%	100%	99.9%	99.3%	Infection-induced acute-onset axonal neuropathy, 621333
REEP1	87.5%	87.5%	100%	100%	99.7%	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011;Spastic paraplegia 31, autosomal dominant, 610250;?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751
RETREG1	91.3%	91.3%	100%	99.9%	99.5%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RNF170	100%	100%	100%	100%	99.9%	Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686

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RTN2	100%	100%	100%	100%	99.2%	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805
SACS	99%	99%	100%	100%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	100%	100%	100%	100%	99.9%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SBF1	100%	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	93.7%	93.7%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCARB2	100%	100%	100%	100%	99.7%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN10A	100%	100%	100%	100%	99.8%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	100%	100%	99.8%	99.4%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548

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SCN9A	100%	100%	100%	100%	99.5%	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%	100%	100%	100%	99.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	95.5%	95.5%	100%	100%	99.7%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEPTIN9	100%	100%	100%	99.9%	98.9%	Amyotrophy, hereditary neuralgic, 162100
SETX	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433
SGPL1	95.5%	95.5%	100%	100%	99.7%	RENI syndrome, 617575
SH3TC2	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353
SIGMAR1	100%	100%	100%	100%	100%	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726;?Amyotrophic lateral sclerosis 16, juvenile, 614373

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SLC12A6	100%	100%	100%	100%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068
SLC25A19	100%	100%	100%	100%	99.8%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A46	100%	100%	100%	100%	99.7%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A6	100%	100%	100%	100%	99.5%	Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903
SLC5A7	100%	100%	100%	99.9%	99.4%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SOD1	100%	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400

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SORD	83%	78.5%	98%	95.9%	93.1%	Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912
SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPAST	94.7%	94.6%	100%	100%	99.5%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	99.6%	99.6%	100%	100%	99.7%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360
SPG7	100%	100%	100%	99.9%	99.3%	Spastic paraplegia 7, autosomal recessive, 607259
SPTAN1	99.2%	98.8%	100%	100%	99.5%	Developmental delay with or without epilepsy, 620540;Developmental and epileptic encephalopathy 5, 613477;Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538;Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528
SPTBN4	100%	100%	100%	99.9%	98.7%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519

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SPTLC1	84.4%	83.8%	100%	100%	99.7%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	89%	89%	100%	100%	99.7%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	91.9%	91.9%	100%	100%	99.9%	
SURF1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SYT2	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040;Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
TBCE	96.7%	96.6%	100%	100%	99.6%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TDP1	87.9%	87.5%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDRKH	100%	100%	100%	99.6%	98.6%	
TECPR2	100%	100%	100%	99.9%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031

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TFG	100%	100%	100%	100%	99.4%	?Spastic paraplegia 57, autosomal recessive, 615658;Hereditary motor and sensory neuropathy, Okinawa type, 604484
TRIM2	96.1%	96.1%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 2R, 615490
TRPV4	100%	100%	100%	99.9%	99.1%	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;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;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500
TTR	100%	100%	100%	100%	99.5%	Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
TUBB2A	100%	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 5, 615763

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TUBB3	100%	100%	100%	100%	99.5%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	100%	100%	100%	100%	99.6%	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%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBA5	94.1%	92.2%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBE3C	94.8%	94.8%	100%	100%	99.6%	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270
UCHL1	100%	100%	100%	100%	99.8%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
UQCRC1	100%	100%	100%	100%	99.5%	Parkinsonism with polyneuropathy, 619279

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VCP	100%	99.6%	100%	100%	99.8%	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	99.6%	98%	100%	100%	99.9%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
VWA1	100%	100%	100%	100%	99.1%	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216
WARS1	91.8%	91.8%	100%	100%	99.6%	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317
WNK1	100%	100%	100%	99.9%	99%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
XRCC1	100%	100%	100%	100%	99.2%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
YARS1	100%	98.5%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323

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ZFYVE26	100%	100%	100%	100%	99.7%	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.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors