NEUROPATHIES GENE PANEL DG 2.17 (149 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AARS	109.2	100.0%	99.7%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
ABCA1	99.2	99.9%	98.7%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCD1	95.7	77.7%	74.9%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABHD12	96.9	100.0%	99.5%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
AIFM1	92.9	99.7%	96.5%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
AMACR	168.4	100.0%	100.0%	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
APTX	99.2	94.5%	91.6%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARHGEF10	126.8	99.8%	98.4%	?Slowed nerve conduction velocity, AD, 608236
ARSA	154.9	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ATAD3A	100.8	93.8%	88.8%	Harel-Yoon syndrome, 617183
ATL1	136.1	100.0%	99.4%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	115.9	99.7%	97.4%	Neuropathy, hereditary sensory, type IF, 615632
ATP1A1	114.1	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP7A	109.4	99.7%	97.2%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
BAG3	189.4	100.0%	99.9%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	166.3	100.0%	99.9%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290

BSCL2	112.9	100.0%	99.9%	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	112.4	100.0%	99.8%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
CCT5	123.2	100.0%	99.1%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHCHD10	28.6	67.7%	41.5%	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CNTNAP1	170.0	100.0%	99.4%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
COA7	132.3	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COX6A1	160.6	100.0%	100.0%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CTDP1	141.7	96.2%	88.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CYP27A1	184.4	100.0%	99.8%	Cerebrotendinous xanthomatosis, 213700
DCAF8	118.2	100.0%	99.5%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCTN1	121.1	100.0%	99.5%	Perry syndrome, 168605 Neuropathy, distal hereditary motor, type VIIB, 607641
DCTN2	94.2	100.0%	99.7%	No OMIM Disease ID
DHTKD1	127.4	99.9%	99.0%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAH10	132.6	99.9%	99.4%	No OMIM Disease ID
DNAJB2	129.2	100.0%	100.0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	138.1	99.5%	96.5%	No OMIM Disease ID
DNM2	134.2	99.8%	97.7%	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	122.3	99.4%	98.7%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DRP2	86.7	98.7%	95.0%	No OMIM Disease ID
DST	145.3	99.9%	99.1%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DYNC1H1	149.0	100.0%	99.8%	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228

EGR2	140.2	100.0%	100.0%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
ELP1	118.4	99.9%	98.3%	Dysautonomia, familial, 223900
EXOSC8	86.9	98.9%	88.8%	Pontocerebellar hypoplasia, type 1C, 616081
FAM126A	124.1	100.0%	98.9%	Leukodystrophy, hypomyelinating, 5, 610532
FBLN5	103.0	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	162.9	99.6%	98.3%	Neuronopathy, distal hereditary motor, type IID, 615575
FGD4	105.3	99.8%	98.3%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	155.0	100.0%	99.7%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FLVCR1	154.8	100.0%	99.4%	Ataxia, posterior column, with retinitis pigmentosa, 609033
GALC	103.0	99.8%	98.1%	Krabbe disease, 245200
GAN	147.5	100.0%	99.6%	Giant axonal neuropathy-1, 256850
GARS	128.2	100.0%	99.6%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GBE1	152.5	100.0%	99.5%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GDAP1	151.2	99.9%	98.7%	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	161.4	100.0%	100.0%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB3	245.5	100.0%	100.0%	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	74.4	99.4%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GNB4	148.6	100.0%	100.0%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
HADHB	76.9	96.0%	83.7%	Trifunctional protein deficiency, 609015

HARS	142.4	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HINT1	65.5	92.7%	79.4%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	123.7	100.0%	99.6%	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	102.8	100.0%	99.0%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HOXD10	168.0	100.0%	100.0%	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HSPB1	77.7	98.0%	92.7%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	238.7	100.0%	100.0%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	235.6	100.0%	100.0%	Neuropathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
IFRD1	138.0	100.0%	98.9%	No OMIM Disease ID
IGHMBP2	117.7	99.9%	98.2%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
INF2	112.4	86.1%	84.3%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
KARS	109.9	100.0%	99.3%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KIF1A	125.3	99.8%	98.2%	Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	144.0	100.0%	99.7%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210
KIF5A	121.9	100.0%	100.0%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
LITAF	109.3	95.2%	91.2%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	118.2	98.3%	93.2%	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	156.9	100.0%	99.9%	No OMIM Disease ID
LRSAM1	145.8	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MARS	106.2	99.9%	98.8%	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MCM3AP	136.4	100.0%	99.4%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MED25	148.0	100.0%	99.9%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MFN2	130.9	100.0%	99.9%	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MME	111.2	99.8%	97.8%	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MORC2	132.4	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPV17	93.2	100.0%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	135.9	100.0%	99.0%	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	99.0	99.9%	98.2%	Charcot-Marie-Tooth disease, type 4B1, 601382
MYH14	120.4	99.4%	96.3%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYO1A	107.8	100.0%	99.9%	No OMIM Disease ID
NAGLU	130.5	98.5%	95.6%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NDRG1	122.5	100.0%	100.0%	Charcot-Marie-Tooth disease, type 4D, 601455
NEFH	121.3	99.5%	98.0%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	205.5	99.9%	97.9%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684

NGF	211.6	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NIPA1	177.0	100.0%	100.0%	Spastic paraplegia 6, autosomal dominant, 600363
NTRK1	144.9	100.0%	99.7%	Medullary thyroid carcinoma, familial, 155240 Insensitivity to pain, congenital, with anhidrosis, 256800
PDK3	109.5	97.4%	94.3%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDYN	131.7	100.0%	100.0%	Spinocerebellar ataxia 23, 610245
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	75.9	100.0%	97.9%	Refsum disease, 266500
PIEZO2	106.9	99.9%	99.4%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PLA2G6	121.0	99.9%	98.6%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLEKHG5	113.7	99.9%	99.0%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	124.4	100.0%	100.0%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100.0	99.0%	95.4%	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	123.1	100.0%	100.0%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PRDM12	153.1	93.7%	91.1%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRPS1	113.2	100.0%	99.9%	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	177.3	100.0%	99.9%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	103.3	100.0%	99.5%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
RAB7A	117.9	100.0%	100.0%	Charcot-Marie-Tooth disease, type 2B, 600882
REEP1	74.4	78.8%	76.7%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
RETREG1	132.5	100.0%	98.6%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SACS	151.1	100.0%	100.0%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SBF1	136.9	99.6%	98.4%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	107.8	99.9%	99.0%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN10A	141.5	100.0%	99.5%	Episodic pain syndrome, familial, 2, 615551
SCN11A	125.5	99.3%	97.2%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	128.2	99.0%	97.6%	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	134.9	100.0%	100.0%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SEPT9	159.1	100.0%	99.9%	Amyotrophy, hereditary neuralgic, 162100
SETX	153.0	100.0%	99.7%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SH3TC2	110.2	100.0%	99.6%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SIGMAR1	162.1	100.0%	100.0%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SLC12A6	120.5	100.0%	100.0%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC25A46	175.2	99.8%	97.2%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC52A2	213.2	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	128.9	100.0%	99.9%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500

SLC5A7	102.5	100.0%	99.9%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SOX10	101.7	100.0%	99.8%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SPG11	118.8	99.9%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPTBN4	117.1	99.9%	99.0%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	108.0	98.4%	91.8%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	146.3	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	134.8	100.0%	99.9%	No OMIM Disease ID
SURF1	89.9	93.5%	89.1%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
SYT2	100.4	99.9%	98.6%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
TBCE	117.0	99.3%	95.6%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TDP1	105.7	100.0%	99.5%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TFG	112.1	97.1%	95.6%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TRIM2	144.0	93.9%	93.4%	Charcot-Marie-Tooth disease, type 2R, 615490
TRPV4	150.4	100.0%	100.0%	Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 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	125.9	94.6%	94.6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TUBB3	135.6	99.9%	99.1%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TWNK	170.3	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3,

				609286 Perrault syndrome 5, 616138
VCP	103.9	100.0%	99.4%	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	126.7	99.9%	98.3%	Pontocerebellar hypoplasia type 1A, 607596
WARS	101.4	99.8%	97.9%	Neuronopathy, distal hereditary motor, type IX, 617721
WNK1	138.8	100.0%	99.6%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
YARS	110.2	100.0%	99.4%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors