# EPILEPSY GENE PANEL DG 2.15 (319 genes)

Releasedate: 31-01-2019

| Gene | Median Coverage | % covered > 10x | % covered > 20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|
| AARS | 124.3 | 100 | 99.6 | Charcot-Marie-Tooth disease, axonal, type 2N, 613287<br>Epileptic encephalopathy, early infantile, 29, 616339 |
| ABAT | 92.7 | 100 | 99.5 | GABA-transaminase deficiency, 613163 |
| ABCC8 | 146.6 | 100 | 99.9 | Diabetes mellitus, noninsulin-dependent, 125853<br>Diabetes mellitus, permanent neonatal, 606176<br>Diabetes mellitus, transient neonatal 2, 610374<br>Hyperinsulinemic hypoglycemia, familial, 1, 256450<br>Hypoglycemia of infancy, leucine-sensitive, 240800 |
| ACTB | 129 | 99.1 | 94.2 | ?Dystonia, juvenile-onset, 607371<br>Baraitser-Winter syndrome 1, 243310 |
| ACY1 | 132.8 | 99.9 | 98.3 | Aminoacylase 1 deficiency, 609924 |
| ADSL | 183.6 | 99.2 | 99.1 | Adenylosuccinase deficiency, 103050 |
| ALDH7A1 | 77.1 | 93.7 | 85.4 | Epilepsy, pyridoxine-dependent, 266100 |
| ALG1 | 50.9 | 53.6 | 48.8 | Congenital disorder of glycosylation, type Ik, 608540 |
| ALG11 | 139.6 | 96.7 | 96 | Congenital disorder of glycosylation, type Ip, 613661 |
| ALG13 | 86.7 | 98.7 | 94.1 | ?Congenital disorder of glycosylation, type Is, 300884<br>Epileptic encephalopathy, early infantile, 36, 300884 |
| ALG3 | 132.9 | 100 | 100 | Congenital disorder of glycosylation, type Id, 601110 |
| ALG6 | 96.4 | 96 | 93.3 | Congenital disorder of glycosylation, type Ic, 603147 |
| AMACR | 157.9 | 100 | 100 | Alpha-methylacyl-CoA racemase deficiency, 614307<br>Bile acid synthesis defect, congenital, 4, 214950 |
| AMPD2 | 135.5 | 99.9 | 99.2 | ?Spastic paraplegia 63, 615686<br>Pontocerebellar hypoplasia, type 9, 615809 |
| AMT | 173.1 | 100 | 100 | Glycine encephalopathy, 605899 |
| ANKRD11 | 96.3 | 97.4 | 94.1 | KBG syndrome, 148050 |
| AP3B2 | 135.1 | 97.5 | 94.2 | Epileptic encephalopathy, early infantile, 48, 617276 |
| APOPT1 | 63.8 | 81.4 | 78.1 | Mitochondrial complex IV deficiency, 220110 |
| ARHGEF9 | 60.2 | 76.4 | 74.4 | Epileptic encephalopathy, early infantile, 8, 300607 |

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| ARID1B | 156.7 | 94.3 | 89.5 | Coffin-Siris syndrome 1, 135900 |
| ARX | 29.1 | 75.8 | 59.5 | Epileptic encephalopathy, early infantile, 1, 308350<br>Hydranencephaly with abnormal genitalia, 300215<br>Lissencephaly, X-linked 2, 300215<br>Mental retardation, X-linked 29 and others, 300419<br>Partington syndrome, 309510<br>Proud syndrome, 300004 |
| ASAH1 | 105.9 | 97.6 | 92.1 | Farber lipogranulomatosis, 228000<br>Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 |
| ASL | 114.4 | 99.9 | 98.8 | Argininosuccinic aciduria, 207900 |
| ASXL3 | 162.9 | 99.8 | 98.9 | Bainbridge-Ropers syndrome, 615485 |
| ATP1A2 | 190.8 | 100 | 99.6 | Alternating hemiplegia of childhood 1, 104290<br>Migraine, familial basilar, 602481<br>Migraine, familial hemiplegic, 2, 602481 |
| ATP1A3 | 177.3 | 100 | 100 | Alternating hemiplegia of childhood 2, 614820<br>CAPOS syndrome, 601338<br>Dystonia-12, 128235 |
| ATP6AP2 | 46.1 | 81.2 | 55.6 | ?Parkinsonism with spasticity, X-linked, 300911<br>Mental retardation, X-linked, syndromic, Hedera type, 300423 |
| ATP7A | 133.2 | 99.7 | 97.8 | Menkes disease, 309400<br>Occipital horn syndrome, 304150<br>Spinal muscular atrophy, distal, X-linked 3, 300489 |
| ATRX | 82.6 | 98.2 | 92.2 | Alpha-thalassemia myelodysplasia syndrome, somatic, 300448<br>Alpha-thalassemia/mental retardation syndrome, 301040<br>Mental retardation-hypotonic facies syndrome, X-linked, 309580 |
| AUTS2 | 110.3 | 96.9 | 95.5 | Mental retardation, autosomal dominant 26, 615834 |
| BOLA3 | 50.1 | 92.3 | 81.7 | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 |
| BRAT1 | 108.5 | 99.8 | 97.4 | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056<br>Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 |
| BTD | 166.6 | 100 | 99.9 | Biotinidase deficiency, 253260 |

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| CACNA1A | 87.8 | 92.7 | 89.1 | Epileptic encephalopathy, early infantile, 42, 617106<br>Episodic ataxia, type 2, 108500<br>Migraine, familial hemiplegic, 1, 141500<br>Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500<br>Spinocerebellar ataxia 6, 183086 |
| CACNA1E | 139.3 | 99.8 | 99.3 | Epileptic encephalopathy, early infantile, 69, 618285 |
| CACNA2D2 | 135.2 | 93.7 | 92.6 | No OMIM phenotype<br>Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154)<br>?Schizophrenia (Purcell (2014) Nature 506, 185) |
| CACNB4 | 106.1 | 96.3 | 94.6 | Episodic ataxia, type 5, 613855<br>{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682<br>{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 |
| CAD | 158.9 | 100 | 99.7 | Epileptic encephalopathy, early infantile, 50, 616457 |
| CASK | 92.3 | 98.7 | 93.7 | FG syndrome 4, 300422<br>Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749<br>Mental retardation, with or without nystagmus, 300422 |
| CDKL5 | 114.4 | 94.9 | 91.8 | Epileptic encephalopathy, early infantile, 2, 300672 |
| CHD2 | 137.7 | 99.3 | 98.5 | Epileptic encephalopathy, childhood-onset, 615369 |
| CHRNA2 | 229.3 | 100 | 100 | Epilepsy, nocturnal frontal lobe, type 4, 610353 |
| CHRNA4 | 142.1 | 96.7 | 95.8 | Epilepsy, nocturnal frontal lobe, 1, 600513<br>{Nicotine addiction, susceptibility to}, 188890 |
| CHRNB2 | 247.5 | 98.4 | 94.6 | Epilepsy, nocturnal frontal lobe, 3, 605375 |
| CLCN4 | 123 | 100 | 99.8 | Raynaud-Claes syndrome, 300114 |
| CLDN16 | 136.3 | 100 | 99.9 | Hypomagnesemia 3, renal, 248250 |
| CLDN19 | 123.7 | 98.2 | 93.7 | Hypomagnesemia 5, renal, with ocular involvement, 248190 |
| CLN3 | 114.9 | 92.5 | 90.7 | Ceroid lipofuscinosis, neuronal, 3, 204200 |
| CLN5 | 146.1 | 98.2 | 92.2 | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| CLN6 | 131.6 | 98.9 | 95.3 | Ceroid lipofuscinosis, neuronal, 6, 601780<br>Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 |
| CLN8 | 163.9 | 83.5 | 83.5 | Ceroid lipofuscinosis, neuronal, 8, 600143<br>Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 |
| CNNM2 | 188.4 | 100 | 99.2 | Hypomagnesemia 6, renal, 613882<br>Hypomagnesemia, seizures, and mental retardation, 616418 |
| CNTN2 | 125.4 | 92.7 | 92.6 | ?Epilepsy, myoclonic, familial adult, 5, 615400 |

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|---|---|---|---|---|
| CNTNAP2 | 148 | 100 | 99.9 | Cortical dysplasia-focal epilepsy syndrome, 610042<br>Pitt-Hopkins like syndrome 1, 610042<br>{Autism susceptibility 15}, 612100 |
| COL4A1 | 92.8 | 97.9 | 94 | ?Retinal arteries, tortuosity of, 180000<br>Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773<br>Brain small vessel disease with or without ocular anomalies, 607595<br>Porencephaly 1, 175780<br>Schizencephaly, 269160<br>{Hemorrhage, intracerebral, susceptibility to}, 614519 |
| COL4A3BP | 121.6 | 98.3 | 92.7 | Mental retardation, autosomal dominant 34, 616351 |
| COQ2 | 89.3 | 96.1 | 93.2 | Coenzyme Q10 deficiency, primary, 1, 607426<br>{Multiple system atrophy, susceptibility to}, 146500 |
| COQ4 | 89.8 | 88.4 | 84.9 | Coenzyme Q10 deficiency, primary, 7, 616276 |
| COQ8A | 134.3 | 100 | 99.1 | Coenzyme Q10 deficiency, primary, 4, 612016 |
| CPA6 | 118.3 | 99.8 | 98.5 | Epilepsy, familial temporal lobe, 5, 614417<br>Febrile seizures, familial, 11, 614418 |
| CPS1 | 143.8 | 100 | 99.8 | Carbamoylphosphate synthetase I deficiency, 237300<br>{Pulmonary hypertension, neonatal, susceptibility to}, 615371<br>{Venoocclusive disease after bone marrow transplantation}, 0 |
| CPT2 | 162.8 | 97.2 | 95.4 | CPT II deficiency, infantile, 600649<br>CPT II deficiency, lethal neonatal, 608836<br>CPT II deficiency, myopathic, stress-induced, 255110<br>{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 |
| CSTB | 82.5 | 97.1 | 82.7 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 |
| CTSD | 163.7 | 98 | 95.3 | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| CTSF | 112.8 | 84.2 | 80.2 | Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 |
| CUL4B | 72.8 | 98 | 88.5 | Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 |
| CUX2 | 85.6 | 99.8 | 97.9 | Epileptic encephalopathy, early infantile, 67, 618141 |
| D2HGDH | 134.5 | 97.5 | 95.2 | D-2-hydroxyglutaric aciduria, 600721 |
| DCX | 113.2 | 100 | 99.7 | Lissencephaly, X-linked, 300067<br>Subcortical laminal heterotopia, X-linked, 300067 |
| DDX3X | 80.5 | 85.9 | 82.1 | Mental retardation, X-linked 102, 300958 |
| DENND5A | 123 | 99.8 | 97.9 | Epileptic encephalopathy, early infantile, 49, 617281 |
| DEPDC5 | 148.3 | 99.8 | 99.3 | Epilepsy, familial focal, with variable foci 1, 604364 |

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| DHDDS | 93.5 | 97.8 | 94.8 | ?Congenital disorder of glycosylation, type 1bb, 613861<br>Developmental delay and seizures with or without movement abnormalities, 617836<br>Retinitis pigmentosa 59, 613861 |
| DLAT | 91.6 | 99.1 | 96 | Pyruvate dehydrogenase E2 deficiency, 245348 |
| DNAJC5 | 200.2 | 100 | 99.9 | Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 |
| DNM1 | 156.7 | 89.3 | 87.5 | Epileptic encephalopathy, early infantile, 31, 616346 |
| DOCK7 | 114.4 | 97.9 | 95.6 | Epileptic encephalopathy, early infantile, 23, 615859 |
| DPAGT1 | 110.7 | 100 | 100 | Congenital disorder of glycosylation, type Ij, 608093<br>Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 |
| DPM1 | 131.1 | 91.7 | 86.7 | Congenital disorder of glycosylation, type Ie, 608799 |
| DPM2 | 102.1 | 100 | 99.4 | Congenital disorder of glycosylation, type Iu, 615042 |
| DPYD | 158.3 | 95.6 | 93.7 | 5-fluorouracil toxicity, 274270<br>Dihydropyrimidine dehydrogenase deficiency, 274270 |
| DPYS | 133.5 | 100 | 99.5 | Dihydropyrimidinuria, 222748 |
| DYNC1H1 | 179.8 | 100 | 99.6 | Charcot-Marie-Tooth disease, axonal, type 20, 614228<br>Mental retardation, autosomal dominant 13, 614563<br>Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 |
| DYRK1A | 159.6 | 100 | 100 | Mental retardation, autosomal dominant 7, 614104 |
| EEF1A2 | 177.7 | 98.8 | 93.8 | Epileptic encephalopathy, early infantile, 33, 616409<br>Mental retardation, autosomal dominant 38, 616393 |
| EFHC1 | 136.3 | 92.2 | 91.5 | {Epilepsy, juvenile absence, susceptibility to, 1}, 607631<br>{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770 |
| EGF | 135.2 | 100 | 99.8 | Hypomagnesemia 4, renal, 611718 |
| EHMT1 | 141.4 | 94 | 92.9 | Kleefstra syndrome 1, 610253 |
| EPM2A | 110.1 | 86.2 | 84 | Epilepsy, progressive myoclonic 2A (Lafora), 254780 |
| EXOSC3 | 88.5 | 97.3 | 89.4 | Pontocerebellar hypoplasia, type 1B, 614678 |
| FA2H | 94.1 | 87.9 | 79.9 | Spastic paraplegia 35, autosomal recessive, 612319 |
| FARS2 | 207.7 | 100 | 100 | Combined oxidative phosphorylation deficiency 14, 614946<br>Spastic paraplegia 77, autosomal recessive, 617046 |
| FGD1 | 85.7 | 92.7 | 86.5 | Aarskog-Scott syndrome, 305400<br>Mental retardation, X-linked syndromic 16, 305400 |
| FGF12 | 95.5 | 99.6 | 96.3 | Epileptic encephalopathy, early infantile, 47, 617166 |

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| FLNA | 138.1 | 100 | 99.5 | ?FG syndrome 2, 300321<br>Cardiac valvular dysplasia, X-linked, 314400<br>Congenital short bowel syndrome, 300048<br>Frontometaphyseal dysplasia 1, 305620<br>Heterotopia, periventricular, 1, 300049<br>Intestinal pseudoobstruction, neuronal, 300048<br>Melnick-Needles syndrome, 309350<br>Otopalatodigital syndrome, type I, 311300<br>Otopalatodigital syndrome, type II, 304120<br>Terminal osseous dysplasia, 300244 |
| FOLR1 | 150.4 | 100 | 100 | Neurodegeneration due to cerebral folate transport deficiency, 613068 |
| FOXG1 | 157.5 | 84.8 | 81 | Rett syndrome, congenital variant, 613454 |
| FOXRED1 | 136.6 | 100 | 99.6 | Mitochondrial complex I deficiency, nuclear type 19, 618241 |
| FRMPD4 | 114.1 | 99.7 | 98 | Mental retardation, X-linked 104, 300983 |
| FRRS1L | 103.1 | 68.3 | 63.4 | Epileptic encephalopathy, early infantile, 37, 616981 |
| FXYD2 | 96.7 | 99.8 | 99.2 | Hypomagnesemia 2, renal, 154020 |
| GABRA1 | 179.5 | 100 | 100 | Epileptic encephalopathy, early infantile, 19, 615744<br>{Epilepsy, childhood absence, susceptibility to, 4}, 611136<br>{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 |
| GABRB3 | 140.5 | 98.1 | 93.7 | Epileptic encephalopathy, early infantile, 43, 617113<br>{Epilepsy, childhood absence, susceptibility to, 5}, 612269 |
| GABRG2 | 137.9 | 91.1 | 90.3 | Epilepsy, generalized, with febrile seizures plus, type 3, 611277<br>Febrile seizures, familial, 8, 611277<br>{Epilepsy, childhood absence, susceptibility to, 2}, 607681 |
| GAMT | 93.5 | 90.9 | 80.7 | Cerebral creatine deficiency syndrome 2, 612736 |
| GCK | 141.4 | 100 | 100 | Diabetes mellitus, noninsulin-dependent, late onset, 125853<br>Diabetes mellitus, permanent neonatal, 606176<br>Hyperinsulinemic hypoglycemia, familial, 3, 602485<br>MODY, type II, 125851 |
| GCSH | 34.2 | 83.1 | 67.8 | ?Glycine encephalopathy, 605899 |
| GLDC | 78.9 | 90.6 | 82.7 | Glycine encephalopathy, 605899 |
| GLRA1 | 123.3 | 100 | 100 | Hyperekplexia 1, 149400 |
| GLRB | 98.6 | 96.6 | 88.8 | Hyperekplexia 2, 614619 |

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| GLUD1 | 74.5 | 94.4 | 84.3 | Hyperinsulinism-hyperammonemia syndrome, 606762 |
| GNAO1 | 167.8 | 93.8 | 93.8 | Epileptic encephalopathy, early infantile, 17, 615473<br>Neurodevelopmental disorder with involuntary movements, 617493 |
| GOSR2 | 127.2 | 95.9 | 95 | Epilepsy, progressive myoclonic 6, 614018 |
| GPC3 | 85.1 | 98.6 | 92.6 | Simpson-Golabi-Behmel syndrome, type 1, 312870<br>Wilms tumor, somatic, 194070 |
| GPHN | 167.2 | 98.4 | 96.9 | Molybdenum cofactor deficiency C, 615501 |
| GRIA3 | 98.2 | 99.3 | 94.7 | Mental retardation, X-linked 94, 300699 |
| GRIN1 | 150.7 | 100 | 99.5 | Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254<br>Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 |
| GRIN2A | 159.1 | 100 | 100 | Epilepsy, focal, with speech disorder and with or without mental retardation, 245570 |
| GRIN2B | 189.4 | 99.9 | 99.3 | Epileptic encephalopathy, early infantile, 27, 616139<br>Mental retardation, autosomal dominant 6, 613970 |
| GRIN2D | 73.1 | 69.1 | 62.1 | Epileptic encephalopathy, early infantile, 46, 617162 |
| GRN | 184.5 | 100 | 100 | Aphasia, primary progressive, 607485<br>Ceroid lipofuscinosis, neuronal, 11, 614706<br>Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 |
| HADH | 110.8 | 98 | 95.1 | 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530<br>Hyperinsulinemic hypoglycemia, familial, 4, 609975 |
| HCFC1 | 105.8 | 99.3 | 96.1 | Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type ), 309541 |
| HCN1 | 122.4 | 99.9 | 97.8 | Epileptic encephalopathy, early infantile, 24, 615871 |
| HLCS | 172.8 | 100 | 100 | Holocarboxylase synthetase deficiency, 253270 |
| HNRNPU | 126.6 | 99.3 | 97.9 | Epileptic encephalopathy, early infantile, 54, 617391 |
| HSD17B10 | 117.1 | 100 | 99.2 | HSD10 mitochondrial disease, 300438 |
| HSD17B4 | 95.1 | 93.9 | 90.8 | D-bifunctional protein deficiency, 261515<br>Perrault syndrome 1, 233400 |
| ICK | 131.6 | 99.9 | 98.2 | Endocrine-cerebroosteodysplasia, 612651<br>{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 |
| IDH2 | 103.5 | 99.6 | 96.9 | D-2-hydroxyglutaric aciduria 2, 613657 |
| IER3IP1 | 73 | 93.2 | 82.2 | Microcephaly, epilepsy, and diabetes syndrome, 614231 |

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| IFIH1 | 113.5 | 99.6 | 97.1 | Aicardi-Goutieres syndrome 7, 615846<br>Singleton-Merten syndrome 1, 182250 |
| IQSEC2 | 61.2 | 92.1 | 82.5 | Mental retardation, X-linked 1/78, 309530 |
| ITPA | 120.2 | 100 | 100 | Epileptic encephalopathy, early infantile, 35, 616647<br>[Inosine triphosphatase deficiency], 613850 |
| JAM3 | 158.6 | 100 | 100 | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 |
| KANSL1 | 172.3 | 99.9 | 99.2 | Koolen-De Vries syndrome, 610443 |
| KATNB1 | 141.7 | 100 | 100 | Lissencephaly 6, with microcephaly, 616212 |
| KCNA1 | 167 | 100 | 99.7 | Episodic ataxia/myokymia syndrome, 160120 |
| KCNA2 | 157.7 | 100 | 99.9 | Epileptic encephalopathy, early infantile, 32, 616366 |
| KCNB1 | 145 | 100 | 99.9 | Epileptic encephalopathy, early infantile, 26, 616056 |
| KCNC1 | 199.2 | 100 | 100 | Epilepsy, progressive myoclonic 7, 616187 |
| KCNH1 | 185.8 | 98.7 | 98.7 | Temple-Baraitser syndrome, 611816<br>Zimmermann-Laband syndrome 1, 135500 |
| KCNJ10 | 213.4 | 89.3 | 89.1 | Enlarged vestibular aqueduct, digenic, 600791<br>SESAME syndrome, 612780 |
| KCNJ11 | 299.5 | 100 | 100 | Diabetes mellitus, transient neonatal, 3, 610582<br>Diabetes, permanent neonatal, with or without neurologic features, 606176<br>Hyperinsulinemic hypoglycemia, familial, 2, 601820<br>Maturity-onset diabetes of the young, type 13, 616329<br>{Diabetes mellitus, type 2, susceptibility to}, 125853 |
| KCNMA1 | 120.4 | 94.4 | 93.2 | ?Cerebellar atrophy, developmental delay, and seizures, 617643<br>Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 |
| KCNQ2 | 85.4 | 90.1 | 86.5 | Epileptic encephalopathy, early infantile, 7, 613720<br>Myokymia, 121200<br>Seizures, benign neonatal, 1, 121200 |
| KCNQ3 | 110.7 | 98.9 | 95.5 | Seizures, benign neonatal, 2, 121201 |
| KCNT1 | 112 | 95.3 | 92.3 | Epilepsy, nocturnal frontal lobe, 5, 615005<br>Epileptic encephalopathy, early infantile, 14, 614959 |
| KCTD7 | 166.7 | 95 | 95 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 |
| KDM5C | 112.6 | 97.9 | 95.1 | Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 |
| KPTN | 112.1 | 100 | 99.9 | Mental retardation, autosomal recessive 41, 615637 |
| LAMB1 | 169.8 | 100 | 99.6 | Lissencephaly 5, 615191 |

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| LGI1 | 163.8 | 97.8 | 94.8 | Epilepsy, familial temporal lobe, 1, 600512 |
| LIAS | 133.7 | 99.5 | 97.1 | Hyperglycinemia, lactic acidosis, and seizures, 614462 |
| MBD5 | 196.2 | 99.9 | 99.6 | Mental retardation, autosomal dominant 1, 156200 |
| MECP2 | 87.3 | 99.1 | 93.1 | Encephalopathy, neonatal severe, 300673<br>Mental retardation, X-linked syndromic, Lubs type, 300260<br>Mental retardation, X-linked, syndromic 13, 300055<br>Rett syndrome, 312750<br>Rett syndrome, atypical, 312750<br>Rett syndrome, preserved speech variant, 312750<br>{Autism susceptibility, X-linked 3}, 300496 |
| MED12 | 105.7 | 98 | 94.8 | Lujan-Fryns syndrome, 309520<br>Ohdo syndrome, X-linked, 300895<br>Opitz-Kaveggia syndrome, 305450 |
| MEF2C | 137.7 | 97.9 | 93.5 | Chromosome 5q14.3 deletion syndrome, 613443<br>Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 |
| MFSD8 | 125.1 | 99.9 | 98.4 | Ceroid lipofuscinosis, neuronal, 7, 610951<br>Macular dystrophy with central cone involvement, 616170 |
| MLC1 | 103.4 | 100 | 99.8 | Megalencephalic leukoencephalopathy with subcortical cysts, 604004 |
| MOCS1 | 87.4 | 98.4 | 93 | Molybdenum cofactor deficiency A, 252150 |
| MOCS2 | 139.5 | 99.6 | 99.6 | Molybdenum cofactor deficiency B, 252160 |
| MPDU1 | 111.8 | 100 | 99.7 | Congenital disorder of glycosylation, type If, 609180 |
| MPDZ | 149 | 98.7 | 96.6 | Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 |
| MTHFR | 126.1 | 98.4 | 97.2 | Homocystinuria due to MTHFR deficiency, 236250<br>{Neural tube defects, susceptibility to}, 601634<br>{Schizophrenia, susceptibility to}, 181500<br>{Thromboembolism, susceptibility to}, 188050<br>{Vascular disease, susceptibility to}, 0 |
| MTOR | 140 | 100 | 99.8 | Focal cortical dysplasia, type II, somatic, 607341<br>Smith-Kingsmore syndrome, 616638 |
| MTRR | 139.1 | 100 | 99.2 | Homocystinuria-megaloblastic anemia, cbl E type, 236270<br>{Neural tube defects, folate-sensitive, susceptibility to}, 601634 |

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| NACC1 | 167.7 | 100 | 99.9 | Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 |
| NANS | 106.1 | 100 | 99.9 | Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 |
| NDUFA1 | 166.8 | 100 | 99.6 | Mitochondrial complex I deficiency, nuclear type 12, 301020 |
| NDUFA11 | 86.9 | 99.5 | 95.8 | Mitochondrial complex I deficiency, nuclear type 14, 618236 |
| NDUFAF1 | 115.6 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 11, 618234 |
| NDUFAF2 | 58.6 | 85.7 | 70.9 | Mitochondrial complex I deficiency, nuclear type 10, 618233 |
| NDUFAF3 | 120.8 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 18, 618240 |
| NDUFAF4 | 79.4 | 98.9 | 91.8 | Mitochondrial complex I deficiency, nuclear type 15, 618237 |
| NDUFAF5 | 95.7 | 98.8 | 94.5 | Mitochondrial complex I deficiency, nuclear type 16, 616238 |
| NDUFB3 | 22.6 | 91.9 | 59.2 | Mitochondrial complex I deficiency, nuclear type 25, 618246 |
| NDUFB9 | 120.1 | 99.8 | 97.4 | ?Mitochondrial complex I deficiency, nuclear type 24, 618245 |
| NDUFS1 | 132.2 | 99.8 | 98.6 | Mitochondrial complex I deficiency, nuclear type 5, 618226 |
| NDUFS2 | 117.8 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 6, 618228 |
| NDUFS3 | 142.4 | 90.7 | 90.6 | Mitochondrial complex I deficiency, nuclear type 8, 618230 |
| NDUFS4 | 147.3 | 100 | 99.1 | Mitochondrial complex I deficiency, nuclear type 1, 252010 |
| NDUFS6 | 119.1 | 99.9 | 99.4 | Mitochondrial complex I deficiency, nuclear type 9, 618232 |
| NDUFV1 | 136.7 | 99.7 | 97.8 | Mitochondrial complex I deficiency, nuclear type 4, 618225 |
| NDUFV2 | 69.5 | 78.7 | 53.9 | Mitochondrial complex I deficiency, nuclear type 7, 618229 |
| NECAP1 | 116.9 | 100 | 99.9 | ?Epileptic encephalopathy, early infantile, 21, 615833 |
| NEDD4L | 105.2 | 71.7 | 70.5 | Periventricular nodular heterotopia 7, 617201 |
| NEU1 | 148.1 | 99.4 | 97.1 | Sialidosis, type I, 256550<br>Sialidosis, type II, 256550 |
| NEXMIF | 139.2 | 99.9 | 99 | Mental retardation, X-linked 98, 300912 |
| NGLY1 | 128 | 100 | 99.5 | Congenital disorder of deglycosylation, 615273 |
| NHLRC1 | 174.2 | 100 | 100 | Epilepsy, progressive myoclonic 2B (Lafora), 254780 |
| NPRL2 | 168.5 | 100 | 100 | Epilepsy, familial focal, with variable foci 2, 617116 |
| NPRL3 | 127.5 | 100 | 99.7 | Epilepsy, familial focal, with variable foci 3, 617118 |
| NRXN1 | 160.9 | 96.8 | 95.7 | Pitt-Hopkins-like syndrome 2, 614325<br>{Schizophrenia, susceptibility to, 17}, 614332 |
| NUBPL | 89.8 | 92.9 | 85.9 | Mitochondrial complex I deficiency, nuclear type 21, 618242 |
| OCLN | 220.7 | 100 | 100 | Pseudo-TORCH syndrome 1, 251290 |

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| OFD1 | 51.5 | 84 | 67.8 | ?Retinitis pigmentosa 23, 300424<br>Joubert syndrome 10, 300804<br>Orofaciodigital syndrome I, 311200<br>Simpson-Golabi-Behmel syndrome, type 2, 300209 |
| OPHN1 | 89 | 99.1 | 96.2 | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 |
| PAFAH1B1 | 105.2 | 89.1 | 81.4 | Lissencephaly 1, 607432<br>Subcortical laminar heterotopia, 607432 |
| PAK3 | 82.8 | 97.6 | 91.8 | Mental retardation, X-linked 30/47, 300558 |
| PC | 149.3 | 97.7 | 94.6 | Pyruvate carboxylase deficiency, 266150 |
| PCDH19 | 224.1 | 100 | 99.3 | Epileptic encephalopathy, early infantile, 9, 300088 |
| PDHA1 | 109.8 | 98.1 | 92.1 | Pyruvate dehydrogenase E1-alpha deficiency, 312170 |
| PDHB | 133 | 99.3 | 96.8 | Pyruvate dehydrogenase E1-beta deficiency, 614111 |
| PDP1 | 209.6 | 100 | 100 | Pyruvate dehydrogenase phosphatase deficiency, 608782 |
| PDX1 | 35.4 | 89 | 72.1 | MODY, type IV, 606392<br>Pancreatic agenesis 1, 260370<br>{Diabetes mellitus, type II, susceptibility to}, 125853 |
| PET100 | 94.5 | 88.8 | 74.8 | Mitochondrial complex IV deficiency, 220110 |
| PEX1 | 115.8 | 97.7 | 95.4 | Heimler syndrome 1, 234580<br>Peroxisome biogenesis disorder 1A (Zellweger), 214100<br>Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 |
| PEX10 | 111.8 | 96.1 | 90.1 | Peroxisome biogenesis disorder 6A (Zellweger), 614870<br>Peroxisome biogenesis disorder 6B, 614871 |
| PEX12 | 168.3 | 100 | 100 | Peroxisome biogenesis disorder 3A (Zellweger), 614859<br>Peroxisome biogenesis disorder 3B, 266510 |
| PEX13 | 197.6 | 99.8 | 98.7 | Peroxisome biogenesis disorder 11A (Zellweger), 614883<br>Peroxisome biogenesis disorder 11B, 614885 |
| PEX14 | 149 | 99.7 | 97.5 | Peroxisome biogenesis disorder 13A (Zellweger), 614887 |
| PEX16 | 137 | 97.1 | 93.1 | Peroxisome biogenesis disorder 8A (Zellweger), 614876<br>Peroxisome biogenesis disorder 8B, 614877 |
| PEX19 | 92.9 | 99.9 | 99.2 | Peroxisome biogenesis disorder 12A (Zellweger), 614886 |
| PEX26 | 76.4 | 100 | 99.8 | Peroxisome biogenesis disorder 7A (Zellweger), 614872<br>Peroxisome biogenesis disorder 7B, 614873 |
| PEX3 | 98.1 | 99.1 | 94.3 | ?Peroxisome biogenesis disorder 10B, 617370<br>Peroxisome biogenesis disorder 10A (Zellweger), 614882 |

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| PEX5 | 111.7 | 99.9 | 98.3 | Peroxisome biogenesis disorder 2A (Zellweger), 214110<br>Peroxisome biogenesis disorder 2B, 202370<br>Rhizomelic chondrodysplasia punctata, type 5, 616716 |
| PEX6 | 94.5 | 90.4 | 86.1 | Heimler syndrome 2, 616617<br>Peroxisome biogenesis disorder 4A (Zellweger), 614862<br>Peroxisome biogenesis disorder 4B, 614863 |
| PGAP3 | 72.8 | 62.5 | 58 | Hyperphosphatasia with mental retardation syndrome 4, 615716 |
| PHF6 | 62.8 | 92.6 | 83.7 | Borjeson-Forssman-Lehmann syndrome, 301900 |
| PHGDH | 115.6 | 100 | 99.8 | Neu-Laxova syndrome 1, 256520<br>Phosphoglycerate dehydrogenase deficiency, 601815 |
| PIGA | 90.5 | 90.4 | 81.3 | Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868<br>Paroxysmal nocturnal hemoglobinuria, somatic, 300818 |
| PIGN | 111.3 | 92.6 | 87.1 | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| PIGO | 147 | 100 | 99.9 | Hyperphosphatasia with mental retardation syndrome 2, 614749 |
| PIGP | 101.4 | 91.6 | 83.8 | ?Epileptic encephalopathy, early infantile, 55, 617599 |
| PIGT | 171.3 | 98.1 | 98 | ?Paroxysmal nocturnal hemoglobinuria 2, 615399<br>Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 |
| PLA2G6 | 117.5 | 99.9 | 98.4 | Infantile neuroaxonal dystrophy 1, 256600<br>Neurodegeneration with brain iron accumulation 2B, 610217<br>Parkinson disease 14, autosomal recessive, 612953 |
| PLCB1 | 142.8 | 100 | 99.7 | Epileptic encephalopathy, early infantile, 12, 613722 |
| PLP1 | 129.2 | 100 | 99.4 | Pelizaeus-Merzbacher disease, 312080<br>Spastic paraplegia 2, X-linked, 312920 |
| PLPBP | 112.6 | 99.1 | 92.6 | Epilepsy, early-onset, vitamin B6-dependent, 617290 |
| PMM2 | 141.1 | 99.9 | 99.4 | Congenital disorder of glycosylation, type Ia, 212065 |
| PNKP | 93 | 99.8 | 97.7 | Ataxia-oculomotor apraxia 4, 616267<br>Microcephaly, seizures, and developmental delay, 613402 |
| PNPO | 66.4 | 100 | 98.3 | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 |

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|---|---|---|---|---|
| POLG | 114.4 | 100 | 99.5 | Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700<br>Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662<br>Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459<br>Progressive external ophthalmoplegia, autosomal dominant 1, 157640<br>Progressive external ophthalmoplegia, autosomal recessive 1, 258450 |
| PPP2R1A | 134 | 91.7 | 91.6 | Mental retardation, autosomal dominant 36, 616362 |
| PPT1 | 144.5 | 90 | 87.3 | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| PQBP1 | 186.1 | 100 | 100 | Renpenning syndrome, 309500 |
| PRF1 | 122.5 | 91.2 | 90.8 | Aplastic anemia, 609135<br>Hemophagocytic lymphohistiocytosis, familial, 2, 603553<br>Lymphoma, non-Hodgkin, 605027 |
| PRICKLE1 | 117.3 | 100 | 100 | Epilepsy, progressive myoclonic 1B, 612437 |
| PRRT2 | 78.9 | 99.9 | 98.4 | Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066<br>Episodic kinesigenic dyskinesia 1, 128200<br>Seizures, benign familial infantile, 2, 605751 |
| PSAP | 114.4 | 99.9 | 99 | Combined SAP deficiency, 611721<br>Gaucher disease, atypical, 610539<br>Krabbe disease, atypical, 611722<br>Metachromatic leukodystrophy due to SAP-b deficiency, 249900 |
| PUM1 | 158 | 100 | 99.9 | Spinocerebellar ataxia 47, 617931 |
| PURA | 121.8 | 94.5 | 87.2 | Mental retardation, autosomal dominant 31, 616158 |
| PYCR2 | 127.6 | 100 | 97.6 | Leukodystrophy, hypomyelinating, 10, 616420 |
| QARS | 166.6 | 100 | 100 | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 |
| RAB39B | 113 | 100 | 99.7 | ?Waisman syndrome, 311510<br>Mental retardation, X-linked 72, 300271 |
| RARS2 | 107.2 | 100 | 99.1 | Pontocerebellar hypoplasia, type 6, 611523 |
| RNASEH2A | 142.1 | 100 | 99.9 | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 103.8 | 93.2 | 87.5 | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 209.2 | 100 | 99.9 | Aicardi-Goutieres syndrome 3, 610329 |
| ROGDI | 112.2 | 97.9 | 95.3 | Kohlschutter-Tonz syndrome, 226750 |
| RPS6KA3 | 79.3 | 94.2 | 83.3 | Coffin-Lowry syndrome, 303600<br>Mental retardation, X-linked 19, 300844 |

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|---|---|---|---|---|
| RRM2B | 128.6 | 99.7 | 97.5 | Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075<br>Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075<br>Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 |
| SAMHD1 | 127.9 | 99.6 | 96.6 | ?Chilblain lupus 2, 614415<br>Aicardi-Goutieres syndrome 5, 612952 |
| SCARB2 | 121.2 | 100 | 99.9 | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 |
| SCN1A | 135.2 | 99.6 | 98 | Epilepsy, generalized, with febrile seizures plus, type 2, 604403<br>Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208<br>Febrile seizures, familial, 3A, 604403<br>Migraine, familial hemiplegic, 3, 609634 |
| SCN1B | 168.3 | 97.1 | 96.1 | Atrial fibrillation, familial, 13, 615377<br>Brugada syndrome 5, 612838<br>Cardiac conduction defect, nonspecific, 612838<br>Epilepsy, generalized, with febrile seizures plus, type 1, 604233<br>Epileptic encephalopathy, early infantile, 52, 617350 |
| SCN2A | 156.7 | 99 | 96.4 | Epileptic encephalopathy, early infantile, 11, 613721<br>Seizures, benign familial infantile, 3, 607745 |
| SCN8A | 198.3 | 100 | 99.7 | ?Cognitive impairment with or without cerebellar ataxia, 614306<br>Epileptic encephalopathy, early infantile, 13, 614558<br>Seizures, benign familial infantile, 5, 617080 |
| SEPSECS | 159.3 | 100 | 100 | Pontocerebellar hypoplasia type 2D, 613811 |
| SIK1 | 85.6 | 97 | 92.4 | Epileptic encephalopathy, early infantile, 30, 616341 |
| SLC12A5 | 125.9 | 85.4 | 81.6 | Epileptic encephalopathy, early infantile, 34, 616645<br>{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 |
| SLC13A5 | 164.1 | 100 | 100 | Epileptic encephalopathy, early infantile, 25, 615905 |
| SLC16A1 | 157.1 | 99.9 | 98.8 | Erythrocyte lactate transporter defect, 245340<br>Hyperinsulinemic hypoglycemia, familial, 7, 610021<br>Monocarboxylate transporter 1 deficiency, 616095 |
| SLC19A3 | 186.4 | 100 | 99.9 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 |
| SLC1A2 | 128 | 99.9 | 99.4 | Epileptic encephalopathy, early infantile, 41, 617105 |

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| SLC25A1 | 71 | 92.2 | 87 | ?Myasthenic syndrome, congenital, 23, presynaptic, 618197<br>Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 |
| SLC25A15 | 192.5 | 98.8 | 95 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| SLC25A22 | 108.7 | 99.5 | 96.9 | Epileptic encephalopathy, early infantile, 3, 609304 |
| SLC2A1 | 190.1 | 92.9 | 92.8 | Dystonia 9, 601042<br>GLUT1 deficiency syndrome 1, infantile onset, severe, 606777<br>GLUT1 deficiency syndrome 2, childhood onset, 612126<br>Stomatin-deficient cryohydrocytosis with neurologic defects, 608885<br>{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 |
| SLC35A2 | 108.8 | 99.7 | 96.8 | Congenital disorder of glycosylation, type IIm, 300896 |
| SLC6A1 | 143.6 | 100 | 100 | Myoclonic-atonic epilepsy, 616421 |
| SLC6A8 | 56.5 | 89.8 | 79.1 | Cerebral creatine deficiency syndrome 1, 300352 |
| SLC9A6 | 104.2 | 97.6 | 91.3 | Mental retardation, X-linked syndromic, Christianson type, 300243 |
| SMARCA2 | 113.8 | 95.7 | 93.7 | Nicolaides-Baraitser syndrome, 601358 |
| SMC1A | 99.4 | 99.9 | 98.8 | Cornelia de Lange syndrome 2, 300590 |
| SMS | 67.8 | 88.3 | 73.9 | Mental retardation, X-linked, Snyder-Robinson type, 309583 |
| SNAP25 | 133.7 | 100 | 99.9 | ?Myasthenic syndrome, congenital, 18, 616330 |
| SPTAN1 | 125.5 | 99.1 | 98.6 | Epileptic encephalopathy, early infantile, 5, 613477 |
| ST3GAL3 | 144.5 | 100 | 99.9 | ?Epileptic encephalopathy, early infantile, 15, 615006<br>Mental retardation, autosomal recessive 12, 611090 |
| ST3GAL5 | 121.9 | 84.4 | 84.2 | Salt and pepper developmental regression syndrome, 609056 |
| STX1B | 152.2 | 100 | 98.4 | Generalized epilepsy with febrile seizures plus, type 9, 616172 |
| STXBP1 | 124.5 | 96.8 | 96.8 | Epileptic encephalopathy, early infantile, 4, 612164 |
| SUOX | 212.6 | 100 | 100 | Sulfite oxidase deficiency, 272300 |
| SYN1 | 64.2 | 74 | 63.2 | Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 |
| SYNGAP1 | 141.3 | 98.4 | 98 | Mental retardation, autosomal dominant 5, 612621 |
| SYNJ1 | 127.2 | 99.3 | 96.1 | Epileptic encephalopathy, early infantile, 53, 617389<br>Parkinson disease 20, early-onset, 615530 |
| SYP | 72 | 99.8 | 94 | Mental retardation, X-linked 96, 300802 |
| SZT2 | 149.5 | 99.5 | 99.2 | Epileptic encephalopathy, early infantile, 18, 615476 |

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| TANGO2 | 145.3 | 100 | 100 | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 |
| TBC1D23 | 86 | 95.7 | 91.5 | Pontocerebellar hypoplasia, type 11, 617695 |
| TBC1D24 | 179.2 | 100 | 100 | Deafness , autosomal recessive 86, 614617<br>Deafness, autosomal dominant 65, 616044<br>DOORS syndrome, 220500<br>Epileptic encephalopathy, early infantile, 16, 615338<br>Myoclonic epilepsy, infantile, familial, 605021 |
| TBCD | 152.9 | 95.5 | 92.3 | Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 |
| TBCE | 128 | 99.9 | 98.2 | Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207<br>Hypoparathyroidism-retardation-dysmorphism syndrome, 241410<br>Kenny-Caffey syndrome, type 1, 244460 |
| TCF4 | 128 | 99.9 | 99.5 | Corneal dystrophy, Fuchs endothelial, 3, 613267<br>Pitt-Hopkins syndrome, 610954 |
| TDP2 | 165.1 | 99.9 | 98.8 | Spinocerebellar ataxia, autosomal recessive 23, 616949 |
| TOE1 | 165.1 | 100 | 100 | Pontocerebellar hypoplasia, type 7, 614969 |
| TPP1 | 146.3 | 100 | 100 | Ceroid lipofuscinosis, neuronal, 2, 204500<br>Spinocerebellar ataxia, autosomal recessive 7, 609270 |
| TREX1 | 242.4 | 100 | 100 | Aicardi-Goutieres syndrome 1, dominant and recessive, 225750<br>Chilblain lupus, 610448<br>Vasculopathy, retinal, with cerebral leukodystrophy, 192315<br>{Systemic lupus erythematosus, susceptibility to}, 152700 |
| TRPM6 | 151.1 | 99.8 | 98.7 | Hypomagnesemia 1, intestinal, 602014 |
| TSC1 | 128.8 | 99.8 | 98.8 | Focal cortical dysplasia, type II, somatic, 607341<br>Lymphangioleiomyomatosis, 606690<br>Tuberous sclerosis-1, 191100 |
| TSC2 | 131.2 | 100 | 99 | ?Focal cortical dysplasia, type II, somatic, 607341<br>Lymphangioleiomyomatosis, somatic, 606690<br>Tuberous sclerosis-2, 613254 |
| TSEN15 | 74.2 | 99 | 93.6 | Pontocerebellar hypoplasia, type 2F, 617026 |
| TSEN2 | 123.8 | 100 | 99.8 | Pontocerebellar hypoplasia type 2B, 612389 |
| TSEN54 | 82.9 | 95.9 | 92.9 | ?Pontocerebellar hypoplasia type 5, 610204<br>Pontocerebellar hypoplasia type 2A, 277470 |

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| | | | | Pontocerebellar hypoplasia type 4, 225753 |
| TUBA1A | 113.2 | 99.9 | 97.8 | Lissencephaly 3, 611603 |
| TUBB2A | 109.9 | 96.7 | 95.6 | Cortical dysplasia, complex, with other brain malformations 5, 615763 |
| TUBB2B | 100 | 100 | 100 | Cortical dysplasia, complex, with other brain malformations 7, 610031 |
| TUBB4A | 121.2 | 96 | 95.3 | Dystonia 4, torsion, autosomal dominant, 128101<br>Leukodystrophy, hypomyelinating, 6, 612438 |
| TUBG1 | 164.2 | 100 | 100 | Cortical dysplasia, complex, with other brain malformations 4, 615412 |
| UBA5 | 75.4 | 94.1 | 77.1 | ?Spinocerebellar ataxia, autosomal recessive 24, 617133<br>Epileptic encephalopathy, early infantile, 44, 617132 |
| UBE3A | 89.8 | 97.8 | 91.4 | Angelman syndrome, 105830 |
| VPS53 | 129.2 | 91.4 | 90.4 | Pontocerebellar hypoplasia, type 2E, 615851 |
| WDR26 | 97.5 | 98.3 | 94.6 | Skraban-Deardorff syndrome, 617616 |
| WDR45 | 75 | 97.4 | 90.1 | Neurodegeneration with brain iron accumulation 5, 300894 |
| WWOX | 130.9 | 100 | 99.7 | Epileptic encephalopathy, early infantile, 28, 616211<br>Esophageal squamous cell carcinoma, somatic, 133239<br>Spinocerebellar ataxia, autosomal recessive 12, 614322 |
| XK | 96.8 | 99.9 | 99.1 | McLeod syndrome with or without chronic granulomatous disease, 300842 |
| YWHAG | 226.6 | 100 | 100 | Epileptic encephalopathy, early infantile, 56, 617665 |
| ZEB2 | 157 | 99.8 | 98.8 | Mowat-Wilson syndrome, 235730 |

*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 31st, 2018.*
*This list is accurate for panel version DG 2.15*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*