# EPILEPSY GENE PANEL DG 2.5/2.6

| *Gene name* | *Median coverage* | *% covered > 10x* | *% covered > 20x* | *Associated Phenotype Description and OMIM disease ID* |
|---|---|---|---|---|
| AARS | 116.4 | 100% | 100% | Epileptic encephalopathy, early infantile, 29,616339<br>Charcot-Marie-Tooth disease, axonal, type 2N,613287 |
| ABAT | 86.7 | 100% | 97% | GABA-transaminase deficiency, 613163 |
| ABCC8 | 139.6 | 100% | 100% | Hyperinsulinemic hypoglycemia, familial, 1, 256450 |
| ACTB | 106.7 | 100% | 93% | ?Dystonia,juvenile onset,607371<br>Baraitser-Winter syndrome,243310 |
| ACY1 | 127.7 | 100% | 97% | Aminoacylase 1 deficiency, 609924 |
| ADSL | 175.6 | 100% | 100% | Adenylosuccinase deficiency, 103050 |
| ALDH7A1 | 67.2 | 95% | 85% | Epilepsy, pyridoxine-dependent, 266100 |
| ALG1 | 49.2 | 50% | 45% | Congenital disorder of glycosylation, type Ik,608540 |
| ALG11 | 154.1 | 100% | 100% | Congenital disorder of glycosylation, type Ip,613661 |
| ALG13 | 57.3 | 97% | 89% | Congenital disorder of glycosylation, type Is, 300884 |
| ALG3 | 101.1 | 100% | 99% | Congenital disorder of glycosylation, type Id,601110 |
| ALG6 | 91.8 | 95% | 94% | Congenital disorder of glycosylation, type Ic,603147 |
| AMACR | 132.1 | 100% | 99% | Alpha-methylacyl-CoA racemase deficiency, 614307 |
| AMT | 144.4 | 100% | 100% | Glycine encephalopathy, 605899 |
| APOPT1 | 62.9 | 87% | 82% | Mitochondrial complex IV deficiency, 220110 |
| ARHGEF9 | 64.2 | 100% | 99% | Epileptic encephalopathy, early infantile, 8, 300607 |
| ARX | 18.6 | 62% | 41% | Epileptic encephalopathy, early infantile 1,308350<br>Hydraencephaly with abnormal genitalia,300215<br>Lissencephaly, X-linked 2,300215<br>Mental retardation, X-linked 29,300419<br>Partington syndrome,309510<br>Proud syndrome,300004 |
| ASAH1 | 118.4 | 96% | 87% | Farber lipogranulomatosis, 228000<br>Spinal muscular atrophy with progressive myoclonic epilepsy,159950 |

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| ATP1A2 | 176.3 | 100% | 100% | Migraine, familial hemiplegic, 2, 602481<br>Migraine,familial basilar,602481<br>Alternating hemiplegia of childhood,104290 |
| ATP1A3 | 169.4 | 100% | 100% | Alternating hemiplegia of childhood 2,614820<br>CAPOS syndrome,601338<br>Dystonia-12,128235 |
| ATP6AP2 | 27.9 | 64% | 41% | ?Mental retardation, X-linked, syndromic,Hedera type, 300423<br>?Parkinsonism with spasticity,X-linked,300911 |
| ATP7A | 88.3 | 99% | 93% | Menkes disease, 309400<br>Occipal horn syndrome,304150<br>Spinal muscular atrophy,distal,X-linked 3,300489 |
| ATRX | 54.3 | 96% | 88% | Alpha-thalassemia/mental retardation syndrome, 301040<br>Alpha-thalassemia myelodysplasia syndrome,somatic,300448<br>Mental retardation-hypotonic facies syndrome,X-linked,309580 |
| AUTS2 | 92.9 | 96% | 94% | Mental retardation, autosomal dominant 26,615834 |
| BOLA3 | 55.4 | 91% | 83% | Multiple mitochondrial dysfunctions syndrome 2, 614299 |
| BRAT1 | 88.8 | 98% | 92% | Rigidity and multifocal seizure syndrome,lethal neonatal,614498 |
| BTD | 122.5 | 100% | 99% | Biotinidase deficiency, 253260 |
| CACNA1A | 84.9 | 93% | 87% | 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 |
| CACNA2D2 | 134.7 | 94% | 92% | No OMIM phenotype<br>Epileptic encephalopathy (Pippucci, PLoS One. 2013 Dec 16;8(12):e82154) |
| CASK | 63.1 | 99% | 90% | Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749<br>Mental retardation,with or without nystagmus,300422<br>FG syndrome 4,300422 |
| CDKL5 | 83 | 95% | 91% | Epileptic encephalopathy, early infantile, 2, 300672 |
| CHD2 | 124.9 | 99% | 98% | Epileptic encephalopathy, childhood-onset, 615369 |
| CHRNA2 | 225.5 | 100% | 100% | Epilepsy, nocturnal frontal lobe, type 4, 610353 |
| CHRNA4 | 135.6 | 96% | 96% | Epilepsy, nocturnal frontal lobe, 1, 600513<br>{Nicotine addiction,susceptibility to},188890 |
| CHRNB2 | 249.4 | 100% | 100% | Epilepsy, nocturnal frontal lobe, 3, 605375 |
| CLDN16 | 140 | 100% | 98% | Hypomagnesemia 3, renal, 248250 |

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| CLDN19 | 102.6 | 100% | 94% | Hypomagnesemia 5, renal, with ocular involvement, 248190 |
| CLN3 | 107.2 | 99% | 97% | Ceroid lipofuscinosis, neuronal, 3, 204200 |
| CLN5 | 144.8 | 97% | 91% | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| CLN6 | 119.2 | 100% | 92% | Ceroid lipofuscinosis, neuronal, 6, 601780<br>Ceroid lipofuscinosis,neuronal,Kufs type,adult onset,204300 |
| CLN8 | 206.6 | 100% | 100% | Ceroid lipofuscinosis, neuronal, 8, 600143<br>Ceroid lipofuscinosis, neuronal, 8,Northern epilepsy variant,610003 |
| CNNM2 | 175.1 | 100% | 98% | Hypomagnesemia 6, renal, 613882<br>Hypomagnesemia, seizures, and mental retardation, 616418 |
| CNTN2 | 121.9 | 99% | 99% | ?Epilepsy, familial adult myoclonic, 5, 615400 |
| CNTNAP2 | 130 | 100% | 99% | Cortical dysplasia-focal epilepsy syndrome, 610042<br>Pitt-Hopkins like syndrome 1,610042<br>{Autism susceptibility 15},612100 |
| COL4A1 | 83 | 96% | 89% | ?Retinal arteries,tortuosity of,18000<br>Angiopathy,hereditary,with nephropathy,aneurysms and muscle cramps,611773<br>Brain small vessel disease w/wo ocular anomalies,607595<br>Porencephaly 1,175780 |
| COL4A3BP | 124.9 | 99% | 95% | Mental retardation, autosomal dominant 34,616351 |
| COQ2 | 71.1 | 97% | 91% | Coenzyme Q10 deficiency, primary, 1, 607426<br>{Multiple system atrophy, susceptibility to},146500 |
| COQ4 | 80.5 | 84% | 81% | Coenzyme Q10 deficiency,primary,7,616276 |
| CPA6 | 110.9 | 100% | 100% | Epilepsy, familial temporal lobe, 5, 614417<br>Febrile seizures,familial,11,614418 |
| CPS1 | 148.8 | 100% | 99% | Carbamoylphosphate synthetase I deficiency, 237300<br>{Pulmonary hypertension,neonatal,susceptibility to},615371<br>{Venoocclusive disease after bone marrow transplantation} |
| CPT2 | 142.8 | 96% | 93% | Myopathy due to CPT II deficiency, 255110<br>CPT deficiency,hepatic,type II,600649<br>CPT II deficiency,lethal neonatal,608836<br>{Encephalopathy,acute,infection-induced,4,susceptibility to},614212 |
| CSTB | 103.4 | 100% | 100% | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 |
| CTSD | 152.8 | 98% | 94% | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| CTSF | 91.5 | 84% | 80% | Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 |
| CUL4B | 47.4 | 93% | 84% | Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 |

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| D2HGDH | 116.5 | 97% | 93% | D-2-hydroxyglutaric aciduria, 600721 |
| DCX | 76.6 | 100% | 100% | Lissencephaly, X-linked, 300067<br>Subcortical laminal heteropia, X-linked, 300067 |
| DEPDC5 | 136.8 | 99% | 99% | Epilepsy, familial focal, with variable foci, 604364 |
| DLAT | 85.7 | 99% | 96% | Pyruvate dehydrogenase E2 deficiency, 245348 |
| DNAJC5 | 163.5 | 100% | 99% | Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 |
| DNM1 | 142.4 | 91% | 90% | Epileptic encephalopathy,early infantile,31,616346 |
| DOCK7 | 112.4 | 96% | 94% | Epileptic encephalopathy, early infantile, 23, 615859 |
| DPAGT1 | 112.3 | 100% | 100% | Congenital disorder of glycosylation, type Ij,608093<br>Myasthenic syndrome,congenital,13,with tubular aggregates,614750 |
| DPM1 | 112.1 | 89% | 84% | Congenital disorder of glycosylation, type Ie,608799 |
| DPM2 | 98.3 | 100% | 100% | Congenital disorder of glycosylation, type Iu,615042 |
| DPYD | 166.8 | 97% | 93% | Dihydropyrimidine dehydrogenase deficiency, 274270<br>5-fluorouracil toxicity,274270 |
| DYNC1H1 | 166.1 | 99% | 98% | Charcot-Marie-Tooth disease, axonal, type 20, 614228<br>Mental retardation, autosomal dominant 13, 614563<br>Spinal muscular atrophy, lower extremity-predominant, AD, 158600 |
| DYRK1A | 153.5 | 100% | 99% | Mental retardation, autosomal dominant 7, 614104 |
| EEF1A2 | 176.2 | 100% | 100% | Epileptic encephalopathy,early infantile,33,616409<br>Mental retardation,autosomal dominant 38,616393 |
| EGF | 138.4 | 100% | 100% | Hypomagnesemia 4, renal, 611718 |
| EHMT1 | 130.5 | 99% | 97% | Kleefstra syndrome, 610253 |
| EPM2A | 111.5 | 85% | 82% | Epilepsy, progressive myoclonic 2A (Lafora), 254780 |
| FA2H | 87.6 | 93% | 81% | Spastic paraplegia 35, autosomal recessive, 612319 |
| FARS2 | 190 | 100% | 99% | Combined oxidative phosphorylation deficiency 14, 614946 |
| FASN | 107 | 100% | 98% | No OMIM phenotype<br>Lennox-Gastaut syndrome (Appenzeller (2014) Am J Hum Genet 95,360)<br>Intellectual disability (Najmabadi (2011) Nature 478, 57)<br>Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360) |
| FGD1 | 53.8 | 89% | 77% | Aarskog-Scott syndrome, 305400<br>Mental retardation,X-linked syndromic 16,305400 |

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| FLNA | 85.1 | 99% | 97% | Cardiac valvurar dysplasia,X-linked,314400<br>Congenital short bowel syndrome,300048<br>FG syndrome 2,300321<br>Frontometaphyseal dysplasia,305620<br>Heteropia,periventricular,300049<br>Heteropia,periventricular,ED variant,300537<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 | 139.5 | 100% | 100% | Neurodegeneration due to cerebral folate transport deficiency, 613068 |
| FOXG1 | 88.7 | 85% | 81% | Rett syndrome, congenital variant, 613454 |
| FOXRED1 | 131.8 | 100% | 98% | Leigh syndrome due to mitochondrial complex I deficiency, 256000<br>Mitochondrial complex I deficiency,252010 |
| FXYD2 | 81.7 | 100% | 100% | Hypomagnesemia-2, renal, 154020 |
| GABRA1 | 176.6 | 100% | 100% | Epileptic encephalopathy,early infantile,19,615744<br>{Epilepsy,childhood absense,susceptibility to,4},611136<br>{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 |
| GABRB3 | 143.2 | 95% | 95% | {Epilepsy,childhood absence, susceptibility to, 5},612269<br>Epileptic encephalopathy (Epi4K consortium, Nature. 2013 Sep 12;501(7466):217-21) |
| GABRG2 | 155.9 | 92% | 92% | Epilepsy, generalized, with febrile seizures plus, type 3, 611277<br>Febrile seizures,familial,8,611277<br>{Epilepsy,childhood absence,susceptibility to,2},607681 |
| GAMT | 94.4 | 98% | 91% | Cerebral creatine deficiency syndrome 2, 612736 |
| GCK | 125.7 | 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 | 38.9 | 93% | 61% | Glycine encephalopathy, 605899 |
| GLDC | 77.2 | 87% | 79% | Glycine encephalopathy, 605899 |
| GLRA1 | 120.6 | 100% | 100% | Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400 |
| GLRB | 87.7 | 97% | 90% | Hyperekplexia 2, autosomal recessive, 614619 |
| GLUD1 | 71.2 | 88% | 81% | Hyperinsulinism-hyperammonemia syndrome, 606762 |
| GNAO1 | 173.4 | 100% | 100% | Epileptic encephalopathy, early infantile, 17, 615473 |

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| GOSR2 | 118.6 | 96% | 93% | Epilepsy, progressive myoclonic 6 |
| GPC3 | 72 | 93% | 85% | Simpson-Golabi-Behmel syndrome, type 1, 312870<br>Wilms tumor,somatic,194070 |
| GPHN | 172.9 | 96% | 96% | Molybdenum cofactor deficiency, type C, 252150 |
| GRIA3 | 65.5 | 96% | 85% | Mental retardation, X-linked 94, 300699 |
| GRIN1 | 132.6 | 100% | 98% | Mental retardation, autosomal dominant 8, 614254 |
| GRIN2A | 157.9 | 100% | 100% | Epilepsy with neurodevelopmental defects, 613971 |
| GRIN2B | 184.8 | 99% | 98% | Mental retardation, autosomal dominant 6, 613970<br>Epileptic encephalopathy,early infantile,27,616139 |
| GRN | 171.5 | 100% | 100% | Aphasia,primary progressive,607485<br>Ceroid lipofuscinosis,neuronal,11,614706<br>Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 |
| HADH | 109.6 | 97% | 94% | 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530<br>Hyperinsulinemic hypoglycemia, familial, 4, 609975 |
| HCN1 | 117.4 | 100% | 100% | Epileptic encephalopathy, early infantile, 24, 615871 |
| HDAC4 | 94.5 | 100% | 99% | Brachydactyly-mental retardation syndrome, 600430 |
| HLCS | 160.7 | 100% | 100% | Holocarboxylase synthetase deficiency, 253270 |
| HNRNPU | 112.4 | 99% | 96% | No OMIM phenotype<br>Developmental delay and intellectual disability (King (2014) Genome Res 24,673)<br>Intellectual disability (Hamdan (2014) PLoS Genet 10,e1004772)<br>Intellectual disability,epilepsy,panhypopituitarism,hypertension & other anomalies (Zhu (2015) Genet Med)<br>Lennox-Gastaut syndrome (Allen (2013) Nature 501,217)<br>Fever-associated epilepsy (Hartmann (2015) Epilepsia 56,e26)<br>Infantile spasms (Du (2014) BMC Med Genet 15,62)<br>Speech delay,seizures and CNS anomalies (Caliebe (2010) Eur J Med Genet 53,179)<br>Seizures (Ballif (2012) Hum Genet 131,145)<br>Epileptic encephalopathy (Mefford (2011) Ann Neurol 70,974)<br>Preaxial polydactyly (Gupta (2014) Am J Med Genet A 164A,186)<br>Intellectual disability & seizures (Thierry (2012) Am J Med Genet A 158A,1633)<br>Thin corpus callosum,psychomotor delay & seizures (Selmer (2012) Eur J Med Genet 55,715) |
| HSD17B10 | 71.2 | 100% | 98% | 17-beta-hydroxysteroid dehydrogenase X deficiency, 300438<br>?Mental retardation,X-linked syndromic 10,300220 |
| HSD17B4 | 100.2 | 93% | 92% | D-bifunctional protein deficiency, 261515<br>Perrault syndrome 1,233400 |

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| IDH2 | 95.2 | 100% | 99% | D-2-hydroxyglutaric aciduria 2, 613657 |
| IER3IP1 | 55.9 | 98% | 85% | Microcephaly, epilepsy, and diabetes syndrome, 614231 |
| IFIH1 | 113.3 | 99% | 96% | Aicardi-Goutieres syndrome 7, 615846<br>Singleton-Merten syndrome 1,182250 |
| IQSEC2 | 37.5 | 87% | 76% | Mental retardation, X-linked 1, 309530 |
| KANSL1 | 70.8 | 96% | 89% | Koolen-De Vries syndrome, 610443 |
| KCNA1 | 170.1 | 100% | 99% | Episodic ataxia/myokymia syndrome, 160120 |
| KCNA2 | 150.1 | 100% | 100% | Epileptic encephalopathy, early infantile, 32,616366 |
| KCNB1 | 124.3 | 100% | 99% | Epileptic encephalopathy, early infantile, 26,616056 |
| KCNC1 | 178.2 | 100% | 100% | Epilepsy, progressive myoclonic 7,616187 |
| KCNH1 | 185.4 | 100% | 99% | Temple-Baraitser syndrome,611816<br>Zimmermann-Laband syndrome 1,135500 |
| KCNJ10 | 199.6 | 100% | 100% | SESAME syndrome, 612780<br>Enlarged vestibular aqueduct,digenic,600791 |
| KCNJ11 | 257 | 100% | 100% | Diabetes mellitus,permanent neonatal,with neurologic features,606176<br>Diabetes mellitus,transient neonatal,3,610582<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 | 133.7 | 100% | 99% | Generalized epilepsy and paroxysmal dyskinesia, 609446 |
| KCNQ2 | 80.5 | 98% | 92% | Epileptic encephalopathy,early infantile,7,613720<br>Myokymia,121200<br>Seizures, benign neonatal, 1, 121200 |
| KCNQ3 | 98.8 | 100% | 94% | Seizures, benign neonatal, type 2, 121201 |
| KCNT1 | 106.6 | 94% | 91% | Epileptic encephalopathy, early infantile, 14, 614959<br>Epilepsy,nocturnal frontal lobe,5,615005 |
| KCTD7 | 113.5 | 95% | 93% | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 |
| KDM5C | 68.6 | 95% | 90% | Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 |
| KIAA2022 | 99.9 | 100% | 98% | Mental retardation,X-linked 98,300912 |
| KPTN | 88.6 | 100% | 99% | Mental retardation, autosomal recessive 41, 615637 |
| LGI1 | 177.2 | 95% | 91% | Epilepsy, familial temporal lobe, 1, 600512 |
| LIAS | 149.8 | 100% | 98% | Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462 |
| MBD5 | 179.2 | 100% | 100% | Mental retardation, autosomal dominant 1, 156200 |

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| MECP2 | 48.4 | 97% | 75% | Ecephalopathy,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>{Autism susceptibility,X-linked 3},300496 |
| MED12 | 64.3 | 95% | 89% | Lujan-Fryns syndrome,309520<br>Ohdo syndrome,X-linked,300895<br>Opitz-Kaveggia syndrome, 305450 |
| MEF2C | 112.8 | 99% | 88% | Chromosome 5q14.3 deletion syndrome,613443<br>Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 |
| MFSD8 | 120.9 | 100% | 98% | Ceroid lipofuscinosis, neuronal, 7, 610951<br>Macular dystrophy with central cone involvement,616170 |
| MOCS1 | 73 | 96% | 91% | Molybdenum cofactor deficiency, type A, 252150 |
| MOCS2 | 145.7 | 99% | 99% | Molybdenum cofactor deficiency, type B, 252150 |
| MPDU1 | 113.6 | 100% | 100% | Congenital disorder of glycosylation, type If |
| MTHFR | 134.9 | 100% | 100% | Homocystinuria due to MTHFR deficiency, 236250<br>{Neural tube defects,susceptibility to},601634<br>{Schizophrenia,susceptibility to},181500<br>{Thromboembolism,susceptibility to},188050 |
| MTOR | 128.5 | 100% | 99% | Smith-Kingsmore syndrome,616638<br>Lennox-Gastaut syndrome (Allen(2013) Nature 501, 217) |
| NDUFA1 | 138.7 | 100% | 97% | Mitochondrial complex I deficiency, 252010 |
| NDUFA11 | 74.8 | 98% | 91% | Mitochondrial complex I deficiency, 252010 |
| NDUFAF1 | 95.1 | 100% | 99% | Mitochondrial complex I deficiency, 252010 |
| NDUFAF2 | 43 | 85% | 47% | Mitochondrial complex I deficiency, 252010<br>Leigh syndrome, 256000 |
| NDUFAF3 | 106.1 | 100% | 100% | Mitochondrial complex I deficiency, 252010 |
| NDUFAF4 | 80.8 | 99% | 94% | Mitochondrial complex I deficiency, 252010 |
| NDUFAF5 | 86.5 | 99% | 91% | Mitochondrial complex I deficiency, 252010 |
| NDUFB3 | 26 | 96% | 56% | Mitochondrial complex I deficiency, 252010 |
| NDUFB9 | 109.1 | 100% | 98% | Mitochondrial complex I deficiency, 252010 |
| NDUFS1 | 134.3 | 97% | 97% | Mitochondrial complex I deficiency, 252010 |
| NDUFS2 | 102.5 | 100% | 99% | Mitochondrial complex I deficiency, 252010 |

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| NDUFS3 | 121.8 | 90% | 90% | Mitochondrial complex I deficiency, 252010 |
| NDUFS4 | 155.3 | 100% | 99% | Mitochondrial complex I deficiency, 252010 |
| NDUFS6 | 124.5 | 100% | 100% | Mitochondrial complex I deficiency, 252010 |
| NDUFV1 | 132.1 | 100% | 97% | Mitochondrial complex I deficiency, 252010 |
| NDUFV2 | 60.6 | 83% | 54% | Mitochondrial complex I deficiency, 252010 |
| NECAP1 | 108.2 | 100% | 100% | ?Epileptic encephalopathy, early infantile,21, 615833 |
| NEDD4L | 137.3 | 100% | 97% | No OMIM phenotype<br>Essential hypertension, association with (Russo (2005) Hypertension 46,488)<br>Epilepsy,photosensitive generalised (Dibbens (2007),Genes Brain Behav 6,750)<br>Infantile spasms (Allen (2013) Nature 501,217) |
| NGLY1 | 117.8 | 100% | 99% | Congenital disorder of glycosylation, type Iv |
| NHLRC1 | 138.8 | 100% | 100% | Epilepsy, progressive myoclonic 2B (Lafora), 254780 |
| NRXN1 | 160 | 99% | 96% | Pitt-Hopkins-like syndrome 2, 614325 |
| NUBPL | 90.3 | 82% | 82% | Mitochondrial complex I deficiency, 252010 |
| OFD1 | 29.6 | 74% | 53% | ?Retinitis pigmentosa 23,300424<br>Joubert syndrome 10,300804<br>Oral-facial-digital syndrome 1, 311200<br>Simpson-Golabi-Behmel syndrome,type 2,300209 |
| OPHN1 | 63.9 | 98% | 94% | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 |
| PAK3 | 53 | 96% | 80% | Mental retardation, X-linked 30/47, 300558 |
| PC | 134.1 | 98% | 94% | Pyruvate carboxylase deficiency, 266150 |
| PCDH19 | 133.3 | 100% | 95% | Epileptic encephalopathy, early infantile, 9, 300088 |
| PDHA1 | 72.7 | 94% | 82% | Pyruvate dehydrogenase E1-alpha deficiency, 312170 |
| PDHB | 127.9 | 97% | 94% | Pyruvate dehydrogenase E1-beta deficiency, 614111 |
| PDP1 | 173.3 | 100% | 100% | Pyruvate dehydrogenase phosphatase deficiency, 608782 |
| PDX1 | 29.1 | 82% | 68% | MODY,type IV,606392<br>Pancreatic agenesis 1,260370<br>{Diabetes mellitus,type II,susceptibility to},125853 |
| PET100 | 110.5 | 89% | 72% | Mitochondrial complex IV deficiency, 220110 |
| PEX1 | 106.9 | 97% | 97% | Peroxisome biogenesis disorder 1A (Zellweger), 214100<br>Peroxisome biogenesis disorder 1B (NALD/IRD),601539 |
| PEX10 | 100.3 | 97% | 87% | Peroxisome biogenesis disorder 6A (Zellweger), 614870<br>Peroxisome biogenesis disorder 6B,614871 |

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| PEX12 | 138.3 | 100% | 100% | Peroxisome biogenesis disorder 3A (Zellweger), 614859<br>Peroxisome biogenesis disorder 3B,266510 |
| PEX13 | 194.6 | 100% | 100% | Peroxisome biogenesis disorder 11A (Zellweger), 614883<br>Peroxisome biogenesis disorder 11B,614885 |
| PEX14 | 109.8 | 100% | 95% | Peroxisome biogenesis disorder 13A (Zellweger), 614887 |
| PEX16 | 114 | 93% | 91% | Peroxisome biogenesis disorder 8A, (Zellweger), 614876<br>Peroxisome biogenesis disorder 8B,614877 |
| PEX19 | 100 | 100% | 100% | Peroxisome biogenesis disorder 12A (Zellweger), 614886 |
| PEX26 | 73.3 | 100% | 98% | Peroxisome biogenesis disorder 7A (Zellweger), 614872<br>Peroxisome biogenesis disorder 7B,614873 |
| PEX3 | 97 | 100% | 99% | Peroxisome biogenesis disorder 10A (Zellweger), 614882 |
| PEX5 | 100.1 | 98% | 94% | Peroxisome biogenesis disorder 2A (Zellweger), 214110<br>Peroxisome biogenesis disorder 2B,202370 |
| PEX6 | 75.8 | 86% | 82% | Peroxisome biogenesis disorder 4A (Zellweger), 614862<br>Peroxisome biogenesis disorder 4B,614863 |
| PGAP3 | 113.5 | 97% | 94% | Hyperphosphatasia with mental retardation syndrome 4, 615716 |
| PHF6 | 46.3 | 87% | 81% | Borjeson-Forssman-Lehmann syndrome, 301900 |
| PHGDH | 119.4 | 100% | 99% | Neu-Laxova syndrome 1,256520<br>Phosphoglycerate dehydrogenase deficiency, 601815 |
| PIGA | 56.5 | 76% | 73% | Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868<br>Paroxysmal nocturnal hemoglobinuria, somatic, 300818 |
| PIGN | 116.9 | 96% | 90% | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| PIGO | 113.4 | 100% | 98% | Hyperphosphatasia with mental retardation syndrome 2, 614749 |
| PIGT | 145.2 | 99% | 98% | ?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398<br>?Paroxysmal nocturnal hemoglobinuria 2, 615399 |
| PLA2G6 | 108.7 | 100% | 98% | Infantile neuroaxonal dystrophy 1, 256600<br>Neurodegeneration with brain iron accumulation 2B,610217<br>Parkinson disease 14,autosomal recessive,612953 |
| PLCB1 | 153.4 | 100% | 100% | Epileptic encephalopathy, early infantile, 12, 613722 |
| PLP1 | 91.3 | 100% | 99% | Pelizaeus-Merzbacher disease, 312080<br>Spastic paraplegia 2,X-linked,312920 |
| PMM2 | 163.1 | 100% | 98% | Congenital disorder of glycosylation, type Ia |
| PNKP | 78.6 | 100% | 95% | Ataxia-oculomotor apraxia,616267<br>Microcephaly, seizures and developmental delay, 613402 |

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|---|---|---|---|---|
| PNPO | 63.8 | 100% | 97% | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 |
| POLG | 107.7 | 100% | 98% | 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 | 124.1 | 93% | 93% | Mental retardation, autosomal dominant 36,616362 |
| PPT1 | 150.6 | 100% | 100% | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| PQBP1 | 97 | 97% | 94% | Renpenning syndrome, 309500 |
| PRICKLE1 | 118.1 | 100% | 100% | Epilepsy, progressive myoclonic 1B, 612437 |
| PRICKLE2 | 114.2 | 100% | 100% | Epilepsy, progressive myoclonic 5,613832 |
| PRRT2 | 62.2 | 100% | 99% | Convulsions,familial infantile,with paroxysmal choreoathetosis,602066<br>Episodic kinesigenic dyskinesia 1, 128200<br>Seizures,benign familial infantile, 2,605751 |
| PURA | 88.9 | 89% | 85% | Mental retardation, autosomal dominant 31 |
| QARS | 145.6 | 100% | 99% | Microcephaly, progressive,seizures, and cerebral and cerebellar atrophy, 615760 |
| RAB39B | 89.2 | 99% | 95% | ?Waisman syndrome,311510<br>Mental retardation, X-linked 72, 300271 |
| RARS2 | 111.7 | 100% | 99% | Pontocerebellar hypoplasia, type 6, 611523 |
| RNASEH2A | 124.1 | 100% | 95% | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 106.7 | 88% | 76% | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 175.9 | 100% | 99% | Aicardi-Goutieres syndrome 3, 610329 |
| ROGDI | 110.8 | 96% | 94% | Kohlschutter-Tonz syndrome, 226750 |
| RPS6KA3 | 55.2 | 88% | 80% | Coffin-Lowry syndrome, 303600<br>Mental retardation,X-linked 19,300844 |
| RRM2B | 133 | 98% | 96% | 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,613077 |
| SAMHD1 | 124.9 | 97% | 94% | Aicardi-Goutieres syndrome 5, 612952<br>Chilblain lupus 2,614415 |
| SCARB2 | 126.6 | 100% | 97% | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 |

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|---|---|---|---|---|
| SCN1A | 151.4 | 99% | 98% | Dravet syndrome, 607208<br>Epilepsy, generalized, with febrile seizures plus, type 2, 604403<br>Febrile seizures,familial,3A,604403<br>Migraine,familial hemiplegic,3,609634 |
| SCN1B | 151.7 | 100% | 96% | Atrial fibrillation,familial,13,615366<br>Brugada syndrome 5,612838<br>Cardiac conduction defect,nonspecific,612838<br>Epilepsy, generalized, with febrile seizures plus, type 1, 604233 |
| SCN2A | 153.2 | 99% | 97% | Epileptic encephalopathy,early infantile,11,613721<br>Seizures, benign familial infantile, 3, 607745 |
| SCN8A | 192.7 | 99% | 97% | ?Cognitive impairment with or without cerebellar ataxia, 614306<br>Epileptic encephalopathy,early infantile,13,614558 |
| SIK1 | 75.5 | 98% | 93% | Epileptic encephalopathy, early infantile, 30,616341 |
| SLC13A5 | 154.3 | 100% | 100% | Epileptic encephalopathy, early infantile, 25, 615905 |
| SLC16A1 | 140.7 | 99% | 96% | Erythrocyte lactate transporter defect, 245340<br>Hyperinsulinemic hypoglycemia,familial,7,610021<br>Monocarboxylate transporter 1 deficiency,616095 |
| SLC19A3 | 168.9 | 100% | 100% | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 |
| SLC25A1 | 64.1 | 97% | 88% | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 |
| SLC25A15 | 212.9 | 98% | 97% | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| SLC25A22 | 91.8 | 100% | 97% | Epileptic encephalopathy, early infantile, 3, 609304 |
| SLC2A1 | 161.8 | 100% | 98% | Dystonia 9,601042<br>GLUT1 deficiency syndrome 1, 606777<br>GLUT1 deficiency syndrome 2, 612126<br>Stomatin-deficient cryohydrocytosis with neurologic defects,608885<br>{Epilepsy, idiopathic generalized,susceptibility,12},614847 |
| SLC35A2 | 66.2 | 97% | 93% | Congenital disorder of glycosylation, type IIm, 300896 |
| SLC6A1 | 139.6 | 100% | 99% | Myoclonic-atonic epilepsy,616421 |
| SLC6A8 | 32.5 | 79% | 66% | Cerebral creatine deficiency syndrome 1, 300352 |
| SLC9A6 | 73.9 | 93% | 83% | Mental retardation, X-linked syndromic, Christianson type, 300243 |
| SMARCA2 | 109.5 | 96% | 94% | Nicolaides-Baraitser syndrome,601358 |
| SMC1A | 67.1 | 99% | 95% | Cornelia de Lange syndrome 2, 300590 |
| SMS | 40.3 | 85% | 61% | Smith-Magenis syndrome, 182290 |

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| SPTAN1 | 116.4 | 99% | 97% | Epileptic encephalopathy, early infantile, 5, 613477 |
| SRPX2 | 51.4 | 98% | 95% | Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 |
| ST3GAL3 | 163.4 | 100% | 100% | Epileptic encephalopathy,early infantile,15,615006<br>Mental retardation, autosomal recessive 12, 611090 |
| ST3GAL5 | 118.3 | 94% | 93% | Amish infantile epilepsy syndrome, 609056 |
| STXBP1 | 124.2 | 100% | 100% | Epileptic encephalopathy, early infantile, 4, 612164 |
| SUOX | 189.8 | 100% | 100% | Sulfite oxidase deficiency, 272300 |
| SYN1 | 44.3 | 70% | 56% | Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 |
| SYNGAP1 | 60.9 | 92% | 79% | Mental retardation, autosomal dominant 5, 612621 |
| SYP | 40 | 93% | 74% | Mental retardation, X-linked 96, 300802 |
| SZT2 | 128.5 | 99% | 99% | Epileptic encephalopathy, early infantile, 18, 615476 |
| TBC1D24 | 149.2 | 100% | 100% | Deafness,autosomal recessive 86,614617<br>Deafness,autosomal dominant 65,616044<br>DOOR syndrome,220500<br>Epileptic encephalopathy,early infantile,16,615338<br>Myoclonic epilepsy, infantile, familial, 605021 |
| TBCE | 147.1 | 99% | 98% | Hypoparathyroidism-retardation-dysmorphism syndrome,241410<br>Kenny-Caffey syndrome-1, 244460 |
| TCF4 | 137.6 | 100% | 100% | Corneal dystrophy,Fuchs endothelial 3,613267<br>Pitt-Hopkins syndrome, 610954 |
| TDP2 | 137.6 | 100% | 98% | No OMIM phenotype<br>Intellectual disability,seizures and ataxia (Gomez-Herreros (2014) Nat Genet 46,516) |
| TPP1 | 124 | 100% | 100% | Ceroid lipofuscinosis, neuronal, 2, 204500<br>Spinocerebellar ataxia,autosomal recessive 7,609270 |
| TREX1 | 214.3 | 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 | 145.2 | 100% | 99% | Hypomagnesemia 1, intestinal, 602014 |
| TSC1 | 127.2 | 99% | 97% | Focal cortical dysplasia, Taylor balloon cell type, 607341<br>Lymphangioleiomyomatosis, 606690<br>Tuberous sclerosis-1, 191100 |
| TUBB2A | 102 | 99% | 95% | Cortical dysplasia, complex, with other brain malformations 5, 615763 |
| TUBG1 | 155.1 | 100% | 100% | Cortical dysplasia,complex,with other brain malformations 4,615412 |

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| UBE3A | 92.8 | 97% | 92% | Angelman syndrome, 105830 |
| WWOX | 122.5 | 100% | 99% | Epileptic encephalopathy, early infantile, 28,616211<br>Esophageal squamous cell carcinoma, somatic,133239<br>Spinocerebellar ataxia,autosomal recessive 12,614322 |
| ZEB2 | 149.6 | 100% | 98% | 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 : April 10th, 2016.*
*This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*