EPILEPSY GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
AARS	91.8	97%	94%	Epileptic encephalopathy, early infantile, 29,616339 Charcot-Marie-Tooth disease, axonal, type 2N,613287
ABAT	62.8	94%	87%	GABA-transaminase deficiency, 613163
ABCC8	86.2	99%	96%	Hyperinsulinemic hypoglycemia, familial, 1, 256450
ACTB	61.8	100%	95%	?Dystonia,juvenile onset,607371 Baraitser-Winter syndrome,243310
ACY1	81.3	100%	96%	Aminoacylase 1 deficiency, 609924
ADSL	120.3	100%	99%	Adenylosuccinase deficiency, 103050
ALDH7A1	69.9	95%	92%	Epilepsy, pyridoxine-dependent, 266100
ALG1	45.6	45%	45%	Congenital disorder of glycosylation, type Ik,608540
ALG11	140.7	100%	100%	Congenital disorder of glycosylation, type Ip,613661
ALG13	111.7	96%	95%	Congenital disorder of glycosylation, type Is, 300884
ALG3	84.6	99%	90%	Congenital disorder of glycosylation, type Id,601110
ALG6	92.2	100%	99%	Congenital disorder of glycosylation, type Ic,603147
AMACR	90.1	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307
AMT	124.2	100%	100%	Glycine encephalopathy, 605899
APOPT1	92.9	87%	87%	Mitochondrial complex IV deficiency, 220110
ARHGEF9	94.8	99%	95%	Epileptic encephalopathy, early infantile, 8, 300607
ARX	60.5	80%	73%	Epileptic encephalopathy, early infantile 1,308350 Hydraencephaly with abnormal genitalia,300215 Mental retardation, X-linked 29,300419 Partington syndrome,309510 Proud syndrome,300004
ASAH1	93.5	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy,159950
ATP1A2	100.9	100%	98%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood,104290

ATP1A3	108.3	100%	98%	Alternating hemiplegia of childhood 2,614820 CAPOS syndrome,601338 Dystonia-12,128235
ATP6AP2	61.2	98%	91%	?Mental retardation, X-linked, syndromic,Hedera type, 300423 ?Parkinsonism with spasticity,X-linked,300911
ATP7A	122.6	100%	100%	Menkes disease, 309400 Occipal horn syndrome,304150 Spinal muscular atrophy,distal,X-linked 3,300489
ATRX	136.1	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome,somatic,300448 Mental retardation-hypotonic facies syndrome,X-linked,309580
AUTS2	116.8	100%	96%	Mental retardation, autosomal dominant 26,615834
BOLA3	53.8	100%	99%	Multiple mitochondrial dysfunctions syndrome 2, 614299
BRAT1	70.2	100%	94%	Rigidity and multifocal seizure syndrome,lethal neonatal,614498
BTD	130.2	100%	100%	Biotinidase deficiency, 253260
CACNA1A	78.9	96%	89%	Episodic ataxia,type 2,108500 Migraine, familial hemiplegic, 1, 141500 Spinocerebellar ataxia 6,183086
CACNA2D2	89.8	93%	91%	No OMIM phenotype Epileptic encephalopathy (Pippucci, PLoS One. 2013 Dec 16;8(12):e82154)
CASK	101	100%	100%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation,with or without nystagmus,300422
CDKL5	129.7	100%	99%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	118	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	119.8	100%	98%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	98.9	98%	95%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction,susceptibility to},188890
CHRNB2	144.2	95%	93%	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	112.8	96%	91%	Hypomagnesemia 3, renal, 248250
CLDN19	71.7	100%	93%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	81.3	97%	95%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	125	97%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	69.9	97%	83%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis,neuronal,Kufs type,adult onset,204300

CLN8	124.6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8,Northern epilepsy variant,610003
CNNM2	136	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	90.7	100%	98%	?Epilepsy, familial adult myoclonic, 5, 615400
CNTNAP2	100	100%	98%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1,610042 {Autism susceptibility 15},612100
COL4A3BP	112.6	100%	100%	Mental retardation, autosomal dominant 34,616351
COQ2	70	97%	85%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to},146500
COQ4	76.9	89%	78%	Coenzyme Q10 deficiency,primary,7,616276
CPA6	125.2	100%	100%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures,familial,11,614418
CPS1	101.8	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension,neonatal,susceptibility to},615371
CPT2	94	95%	91%	Myopathy due to CPT II deficiency, 255110 CPT deficiency,hepatic,type II,600649 CPT II deficiency,lethal neonatal,608836 {Encephalopathy,acute,infection-induced,4,susceptibility to},614212
CSTB	163.5	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	93.6	100%	97%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	103.7	98%	83%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	108.6	99%	98%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	63.9	96%	86%	D-2-hydroxyglutaric aciduria, 600721
DCX	114.5	100%	100%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DEPDC5	106.3	99%	98%	Epilepsy, familial focal, with variable foci, 604364
DLAT	98.3	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	70.6	92%	80%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	76.7	94%	80%	Epileptic encephalopathy,early infantile,31,616346
DOCK7	100.7	100%	99%	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	91.8	100%	95%	Congenital disorder of glycosylation, type Ij,608093 Myasthenic syndrome,congenital,13,with tubular aggregates,614750

DPM1	132.2	90%	90%	Congenital disorder of glycosylation, type Ie,608799
DPM2	87.5	99%	99%	Congenital disorder of glycosylation, type Iu,615042
DPYD	115.7	99%	97%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity,274270
DYNC1H1	112.5	99%	97%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	132.4	99%	98%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	99.6	100%	100%	Epileptic encephalopathy,early infantile,33,616409 Mental retardation,autosomal dominant 38,616393
EGF	108.2	100%	97%	Hypomagnesemia 4, renal, 611718
EHMT1	92.6	96%	93%	Kleefstra syndrome, 610253
EPM2A	68	87%	84%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	55.7	93%	73%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	101.4	100%	95%	Combined oxidative phosphorylation deficiency 14, 614946
FASN	85.8	98%	94%	No OMIM phenotype Lennox-Gastaut syndrome (Appenzeller (2014) Am J Hum Genet 95,360) Intellectual disability (Najmabadi (2011) Nature 478, 57)
FGD1	88.6	99%	95%	Aarskog-Scott syndrome, 305400 Mental retardation,X-linked syndromic 16,305400
FLNA	119.2	100%	99%	Cardiac valvurar dysplasia,X-linked,314400 Congenital short bowel syndrome,300048 FG syndrome 2,300321 Frontometaphyseal dysplasia,305620 Heteropia,periventricular,300049 Heteropia,periventricular,ED variant,300537
FOLR1	83.3	97%	93%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	86.2	84%	78%	Rett syndrome, congenital variant, 613454
FOXRED1	93.6	100%	92%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency,252010
FXYD2	61.4	93%	88%	Hypomagnesemia-2, renal, 154020
GABRA1	123.5	99%	96%	Epileptic encephalopathy,early infantile,19,615744 {Epilepsy,childhood absense,susceptibility to,4},611136

GABRB3	112.6	100%	95%	{Epilepsy,childhood absence, susceptibility to, 5},612269 Epileptic encephalopathy (Epi4K consortium, Nature. 2013 Sep 12;501(7466):217-21)
GABRG2	120.9	99%	92%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures,familial,8,611277 {Epilepsy,childhood absence,susceptibility to,2},607681
GAMT	99.9	98%	93%	Cerebral creatine deficiency syndrome 2, 612736
GCK	85.3	100%	99%	Diabetes mellitus,noninsulin-dependent,late onset,125853 Diabetes mellitus,permanent neonatal,606176 Hyperinsulinemic hypoglycemia,familial,3,602485 MODY, type II, 125851
GCSH	12.4	51%	33%	Glycine encephalopathy, 605899
GLDC	57.6	97%	85%	Glycine encephalopathy, 605899
GLRA1	112.6	100%	99%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	110.9	100%	97%	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	110.2	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	112.3	100%	98%	Epileptic encephalopathy, early infantile, 17, 615473
GOSR2	94.7	97%	93%	Epilepsy, progressive myoclonic 6
GPC3	96	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor,somatic,194070
GPHN	116.8	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GRIA3	99.7	100%	97%	Mental retardation, X-linked 94, 300699
GRIN1	81.7	99%	96%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	134.7	100%	99%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	135.5	99%	98%	Mental retardation, autosomal dominant 6, 613970 Epileptic encephalopathy,early infantile,27,616139
GRN	114.2	100%	99%	Aphasia,primary progressive,607485 Ceroid lipofuscinosis,neuronal,11,614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HADH	76.9	100%	98%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCN1	95.7	100%	99%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	66.1	93%	90%	Brachydactyly-mental retardation syndrome, 600430
HLCS	140.4	100%	100%	Holocarboxylase synthetase deficiency, 253270

HNRNPU	103.6	97%	96%	No OMIM phenotype Developmental delay and intellectual disability (King (2014) Genome Res 24, 673) Infantile spasms (Du (2014) BMC Med Genet 15, 62) Speech delay, seizures & CNS anomalies (Caliebe (2010) Eur J Med Genet 53, 179)
HSD17B10	105.3	99%	92%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation,X-linked syndromic 10,300220
HSD17B4	90	96%	95%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1,233400
IDH2	107.5	100%	99%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	58.5	100%	93%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	127.2	100%	99%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1,182250
IQSEC2	78.8	96%	87%	Mental retardation, X-linked 1, 309530
KANSL1	69.6	96%	86%	Koolen-De Vries syndrome, 610443
KCNA1	102.4	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	208.8	100%	99%	Epileptic encephalopathy, early infantile, 32,616366
KCNB1	150.5	100%	95%	Epileptic encephalopathy, early infantile, 26,616056
KCNC1	137.5	100%	100%	Epilepsy, progressive myoclonic 7,616187
KCNH1	110	100%	99%	Temple-Baraitser syndrome,611816 Zimmermann-Laband syndrome 1,135500
KCNJ10	148.5	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct,digenic,600791
KCNJ11	127.4	100%	100%	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNMA1	84	99%	94%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	70.7	98%	96%	Epileptic encephalopathy,early infantile,7,613720 Myokymia,121200 Seizures, benign neonatal, 1, 121200
KCNQ3	101.8	100%	96%	Seizures, benign neonatal, type 2, 121201
KCNT1	72	96%	92%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy,nocturnal frontal lobe,5,615005
KCTD7	87	71%	68%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	112	100%	100%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534

KPTN	75.9	100%	98%	Mental retardation, autosomal recessive 41, 615637
LGI1	133.6	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	105.9	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
MBD5	143.4	100%	99%	Mental retardation, autosomal dominant 1, 156200
MECP2	163.2	100%	98%	Ecephalopathy,neonatal severe,300673 Mental retardation,X-linked syndromic,Lubs type,300260 Mental retardation,X-linked,syndromic 13,300055 Rett syndrome, 312750 {Autism susceptibility,X-linked 3},300496
MED12	122.4	97%	94%	Lujan-Fryns syndrome,309520 Ohdo syndrome,X-linked,300895 Opitz-Kaveggia syndrome, 305450
MEF2C	105.4	100%	99%	Chromosome 5q14.3 deletion syndrome,613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	105.4	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement,616170
MOCS1	79.9	99%	95%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	112.2	99%	99%	Molybdenum cofactor deficiency, type B, 252150
MPDU1	121.4	100%	100%	Congenital disorder of glycosylation, type If
MTHFR	95.1	100%	98%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects,susceptibility to},601634 {Schizophrenia,susceptibility to},181500 {Thromboembolism,susceptibility to},188050
MTOR	98.6	100%	98%	Smith-Kingsmore syndrome,616638 Lennox-Gastaut syndrome (Allen(2013) Nature 501, 217)
NDUFA1	189.6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	92.3	96%	80%	Mitochondrial complex I deficiency, 252010
NDUFAF1	100.4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	50.7	100%	96%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	127.9	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	66	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	122	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFB3	0.5	0%	0%	Mitochondrial complex I deficiency, 252010

NDUFB9	111	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS1	76.8	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	122.3	100%	96%	Mitochondrial complex I deficiency, 252010
NDUFS3	138.5	91%	90%	Mitochondrial complex I deficiency, 252010
NDUFS4	126.6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS6	118.3	97%	92%	Mitochondrial complex I deficiency, 252010
NDUFV1	62.3	97%	91%	Mitochondrial complex I deficiency, 252010
NDUFV2	100.9	98%	98%	Mitochondrial complex I deficiency, 252010
NECAP1	99.4	100%	100%	?Epileptic encephalopathy, early infantile,21, 615833
NEDD4L	112.6	100%	100%	No OMIM phenotype
NGLY1	107.2	100%	98%	Congenital disorder of glycosylation, type Iv
NHLRC1	107.4	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NRXN1	114.5	99%	97%	Pitt-Hopkins-like syndrome 2, 614325
NUBPL	87.2	100%	100%	Mitochondrial complex I deficiency, 252010
OFD1	68.6	93%	88%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome,type 2,300209
OPHN1	98.8	99%	98%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	100.9	100%	100%	Mental retardation, X-linked 30/47, 300558
PC	93	97%	91%	Pyruvate carboxylase deficiency, 266150
PCDH19	138.2	99%	98%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	116.6	100%	99%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	99.6	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	156.4	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	39.6	100%	94%	MODY,type IV,606392 Pancreatic agenesis 1,260370 {Diabetes mellitus,type II,susceptibility to},125853
PET100	71.8	100%	99%	Mitochondrial complex IV deficiency, 220110
PEX1	118.6	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD),601539
PEX10	73.1	89%	85%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B,614871

PEX12	114	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B,266510
PEX13	128.3	99%	96%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B,614885
PEX14	90.9	100%	99%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	86	92%	83%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B,614877
PEX19	107.5	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	111.4	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B,614873
PEX3	130.7	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	84.9	98%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B,202370
PEX6	89.7	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B,614863
PGAP3	64.7	100%	87%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	137.9	100%	100%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	86.3	100%	99%	Neu-Laxova syndrome 1,256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	134.6	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	102.3	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	107.7	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	123.2	100%	100%	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PLA2G6	76.2	100%	92%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B,610217 Parkinson disease 14,autosomal recessive,612953
PLCB1	107.8	99%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	77.9	100%	94%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2,X-linked,312920
PMM2	85.6	100%	100%	Congenital disorder of glycosylation, type Ia
PNKP	67.7	99%	95%	Ataxia-oculomotor apraxia,616267 Microcephaly, seizures and developmental delay, 613402

PNPO	69.7	100%	90%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	86.5	99%	91%	Mitochondrial DNA depletion syndrome 4A (Alpers type),203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type),613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE),607459 Progressive external ophthalmoplegia, autosomal dominant,1
PPP2R1A	89	93%	90%	Mental retardation, autosomal dominant 36,616362
PPT1	68.4	100%	94%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	121.6	100%	99%	Renpenning syndrome, 309500
PRICKLE1	105.4	100%	98%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	107.1	95%	95%	Epilepsy, progressive myoclonic 5,613832
PRRT2	80.1	100%	100%	Convulsions,familial infantile,with paroxysmal choreoathetosis,602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures,benign familial infantile, 2,605751
PURA	109.8	100%	85%	Mental retardation, autosomal dominant 31
QARS	117.7	99%	99%	Microcephaly, progressive,seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	166.5	100%	100%	?Waisman syndrome,311510 Mental retardation, X-linked 72, 300271
RARS2	82.3	100%	98%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	96.1	99%	94%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	102.3	99%	97%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	133.2	100%	100%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	94.2	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	97.3	100%	99%	Coffin-Lowry syndrome, 303600 Mental retardation,X-linked 19,300844
RRM2B	110	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type),612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal dominant, 5,6130
SAMHD1	112.6	100%	98%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2,614415
SCARB2	97.3	100%	97%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	114.5	100%	99%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures,familial,3A,604403 Migraine,familial hemiplegic,3,609634

SCN1B	109.1	99%	96%	Atrial fibrillation,familial,13,615366 Brugada syndrome 5,612838 Cardiac conduction defect,nonspecific,612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	122	99%	99%	Epileptic encephalopathy,early infantile,11,613721 Seizures, benign familial infantile, 3, 607745
SCN8A	134.9	100%	99%	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy,early infantile,13,614558
SIK1	56.7	94%	77%	Epileptic encephalopathy, early infantile, 30,616341
SLC13A5	82.6	100%	97%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	142.4	100%	100%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia,familial,7,610021 Monocarboxylate transporter 1 deficiency,616095
SLC19A3	107.5	100%	99%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	73.5	83%	80%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	102.9	95%	80%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	73.5	98%	93%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	81.3	100%	100%	Dystonia 9,601042 GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized,susceptibility,12}
SLC35A2	105.8	100%	99%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	87.9	99%	97%	Myoclonic-atonic epilepsy,616421
SLC6A8	7.8	20%	11%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	110.6	100%	97%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	86.4	97%	94%	Nicolaides-Baraitser syndrome,601358
SMC1A	135.7	98%	97%	Cornelia de Lange syndrome 2, 300590
SMS	30.1	87%	64%	Smith-Magenis syndrome, 182290
SPTAN1	95.3	99%	97%	Epileptic encephalopathy, early infantile, 5, 613477
SRPX2	81.6	100%	97%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	120.2	100%	100%	Epileptic encephalopathy,early infantile,15,615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	102.4	93%	92%	Amish infantile epilepsy syndrome, 609056

STXBP1	91	100%	97%	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	174.6	100%	100%	Sulfite oxidase deficiency, 272300
SYN1	69.1	96%	75%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	52.8	92%	80%	Mental retardation, autosomal dominant 5, 612621
SYP	93.4	100%	99%	Mental retardation, X-linked 96, 300802
SZT2	99.9	99%	96%	Epileptic encephalopathy, early infantile, 18, 615476
TBC1D24	109.5	100%	100%	Deafness,autosomal recessive 86,614617 Deafness,autosomal dominant 65,616044 DOOR syndrome,220500 Epileptic encephalopathy,early infantile,16,615338 Myoclonic epilepsy, infantile, familial, 605021
TBCE	109.7	100%	98%	Hypoparathyroidism-retardation-dysmorphism syndrome,241410 Kenny-Caffey syndrome-1, 244460
TCF4	90.8	97%	97%	Corneal dystrophy,Fuchs endothelial 3,613267 Pitt-Hopkins syndrome, 610954
TDP2	132.9	100%	100%	No OMIM phenotype
TPP1	119.3	100%	96%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia,autosomal recessive 7,609270
TREX1	134.4	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus,610448 Vasculopathy,retinal,with cerebral leukodystrophy,192315 {Systemic lupus erythematosus,susceptibility to},152700
TRPM6	112.9	100%	98%	Hypomagnesemia 1, intestinal, 602014
TSC1	89.9	99%	97%	Focal cortical dysplasia, Taylor balloon cell type, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TUBB2A	47.6	100%	93%	Cortical dysplasia, complex, with other brain malformations 5, 615763
UBE3A	101.5	100%	99%	Angelman syndrome, 105830
WWOX	102.5	97%	97%	Epileptic encephalopathy, early infantile, 28,616211 Esophageal squamous cell carcinoma, somatic,133239 Spinocerebellar ataxia,autosomal recessive 12,614322
ZEB2	150.3	100%	99%	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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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