EPILEPSY GENE PANEL DG 2.3.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
AARS	96,6	97%	93%	Epileptic encephalopathy, early infantile, 29,616339 Charcot-Marie-Tooth disease, axonal, type 2N,613287
ABAT	75,3	96%	93%	GABA-transaminase deficiency, 613163
ABCC8	97,3	100%	98%	Hyperinsulinemic hypoglycemia, familial, 1, 256450
ACY1	93,1	99%	97%	Aminoacylase 1 deficiency, 609924
ADSL	133,8	100%	100%	Adenylosuccinase deficiency, 103050
ALDH7A1	78,4	100%	96%	Epilepsy, pyridoxine-dependent, 266100
ALG1	51,6	45%	45%	Congenital disorder of glycosylation, type Ik,608540
ALG11	167,7	100%	100%	Congenital disorder of glycosylation, type Ip,613661
ALG13	61,8	94%	90%	Congenital disorder of glycosylation, type Is, 300884
ALG3	93,4	100%	96%	Congenital disorder of glycosylation, type Id,601110
ALG6	105	100%	100%	Congenital disorder of glycosylation, type Ic,603147
AMACR	99,8	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307
AMT	135,8	100%	100%	Glycine encephalopathy, 605899
APOPT1	95,7	87%	86%	Mitochondrial complex IV deficiency, 220110
ARHGEF9	49,9	97%	88%	Epileptic encephalopathy, early infantile, 8, 300607
ARX	33,7	76%	61%	Epileptic encephalopathy, early infantile 1,308350 Hydraencephaly with abnormal genitalia,300215 Lissencephaly, X-linked 2,300215 Mental retardation, X-linked 29,300419 Partington syndrome,309510 Proud syndrome,300004
ASAH1	104,1	100%	98%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy,159950
ATP1A2	110,2	100%	99%	Migraine, familial hemiplegic, 2, 602481 Migraine,familial basilar,602481 Alternating hemiplegia of childhood,104290

ATP1A3	117,5	100%	99%	Alternating hemiplegia of childhood 2,614820 CAPOS syndrome,601338 Dystonia-12,128235
ATP6AP2	38,3	96%	85%	?Mental retardation, X-linked, syndromic,Hedera type, 300423 ?Parkinsonism with spasticity,X-linked,300911
ATP7A	69,4	100%	99%	Menkes disease, 309400 Occipal horn syndrome,304150 Spinal muscular atrophy,distal,X-linked 3,300489
ATRX	75,1	100%	98%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome,somatic,300448 Mental retardation-hypotonic facies syndrome,X-linked,309580
AUTS2	126,5	100%	99%	Mental retardation, autosomal dominant 26,615834
BOLA3	59,4	100%	92%	Multiple mitochondrial dysfunctions syndrome 2, 614299
BTD	157,3	100%	100%	Biotinidase deficiency, 253260
CACNA1A	81,9	97%	89%	Episodic ataxia,type 2,108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic,1,with progressive cerebellar ataxia,141500 Spinocerebellar ataxia 6,183086
CACNA2D2	96,7	93%	92%	No OMIM phenotype Epileptic encephalopathy (Pippucci, PLoS One. 2013 Dec 16;8(12):e82154)
CASK	53	99%	91%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation,with or without nystagmus,300422 FG syndrome 4,300422
CDKL5	74,1	100%	96%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	128,2	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	126,7	100%	98%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	94,8	100%	97%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction,susceptibility to},188890
CHRNB2	165,3	96%	95%	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	138,3	100%	95%	Hypomagnesemia 3, renal, 248250
CLDN19	85,7	100%	93%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	90,6	97%	97%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	135,9	98%	95%	Ceroid lipofuscinosis, neuronal, 5, 256731

CLN6	75,7	99%	91%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis,neuronal,Kufs type,adult onset,204300
CLN8	139,7	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8,Northern epilepsy variant,610003
CNNM2	152,8	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	93,7	100%	99%	?Epilepsy, familial adult myoclonic, 5, 615400
CNTNAP2	109,8	100%	100%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1,610042 {Autism susceptibility 15},612100
COL4A3BP	112,7	100%	100%	Mental retardation, autosomal dominant 34,616351
COQ2	76,9	97%	85%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to},146500
CPA6	135,7	100%	100%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures,familial,11,614418
CPS1	115,6	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension,neonatal,susceptibility to},615371 {Venoocclusive disease after bone marrow transplantation}
CPT2	105,7	93%	92%	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	186,4	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	98,3	99%	94%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	112,6	99%	87%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	68,3	99%	99%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	72,1	97%	94%	D-2-hydroxyglutaric aciduria, 600721
DCX	66,5	100%	95%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DEPDC5	115,8	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DLAT	106	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	72,9	97%	90%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	86	90%	77%	Epileptic encephalopathy,early infantile,31,616346
DOCK7	110,5	100%	100%	Epileptic encephalopathy, early infantile, 23, 615859

DPAGT1	105,7	100%	99%	Congenital disorder of glycosylation, type Ij,608093 Myasthenic syndrome,congenital,13,with tubular aggregates,614750
DPM1	157,5	90%	90%	Congenital disorder of glycosylation, type Ie,608799
DPM2	80,1	99%	99%	Congenital disorder of glycosylation, type Iu,615042
DPYD	120,9	100%	98%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity,274270
DYNC1H1	127,6	99%	98%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	137,2	99%	99%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	105	100%	98%	Epileptic encephalopathy,early infantile,33,616409 Mental retardation,autosomal dominant 38,616393
EGF	115,5	100%	98%	Hypomagnesemia 4, renal, 611718
EHMT1	104,1	98%	94%	Kleefstra syndrome, 610253
EPM2A	78,8	88%	85%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	63,8	94%	87%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	114,9	100%	96%	Combined oxidative phosphorylation deficiency 14, 614946
FASN	93,2	97%	94%	No OMIM phenotype Lennox-Gastaut syndrome (Appenzeller (2014) Am J Hum Genet 95,360) Intellectual disability (Najmabadi (2011) Nature 478, 57) Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360)
FGD1	48,5	95%	85%	Aarskog-Scott syndrome, 305400 Mental retardation,X-linked syndromic 16,305400
FLNA	65,7	99%	93%	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 Intestinal pseudoobstruction,neuronal,300048 Melnick-Needles syndrome,309350 Otopalatodigital syndrome, type I,311300 Otopalatodigital syndrome, typw II,304120 Terminal osseous dysplasia,300244

FOLR1	90	100%	96%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	92,1	92%	81%	Rett syndrome, congenital variant, 613454
FOXRED1	98,5	100%	98%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency,252010
FXYD2	73,4	99%	80%	Hypomagnesemia-2, renal, 154020
GABBR2	90,4	99%	95%	{Nicotine dependance},188890 Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360)
GABRA1	125,3	99%	94%	Epileptic encephalopathy,early infantile,19,615744 {Epilepsy,childhood absense,susceptibility to,4},611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	116,2	100%	97%	{Epilepsy,childhood absence, susceptibility to, 5},612269 Epileptic encephalopathy (Epi4K consortium, Nature. 2013 Sep 12;501(7466):217-21)
GABRG2	130,8	97%	94%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures,familial,8,611277 {Epilepsy,childhood absence,susceptibility to,2},607681
GAMT	109,2	100%	99%	Cerebral creatine deficiency syndrome 2, 612736
GCK	91,2	100%	100%	Diabetes mellitus,noninsulin-dependent,late onset,125853 Diabetes mellitus,permanent neonatal,606176 Hyperinsulinemic hypoglycemia,familial,3,602485 MODY, type II, 125851
GCSH	13,8	52%	26%	Glycine encephalopathy, 605899
GLDC	61,3	98%	90%	Glycine encephalopathy, 605899
GLRA1	120,8	100%	100%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	128	100%	96%	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	124,8	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	119,9	100%	99%	Epileptic encephalopathy, early infantile, 17, 615473
GOSR2	97,6	97%	97%	Epilepsy, progressive myoclonic 6
GPC3	54,5	99%	96%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor,somatic,194070
GPHN	123,4	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GRIA3	58,3	97%	90%	Mental retardation, X-linked 94, 300699
GRIN1	91	100%	98%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	144,3	99%	98%	Epilepsy with neurodevelopmental defects, 613971

GRIN2B	154,6	99%	98%	Mental retardation, autosomal dominant 6, 613970 Epileptic encephalopathy,early infantile,27,616139
GRN	127,5	100%	100%	Aphasia,primary progressive,607485 Ceroid lipofuscinosis,neuronal,11,614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HADH	91,2	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCN1	103,3	100%	99%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	74,5	95%	90%	Brachydactyly-mental retardation syndrome, 600430
HLCS	152,2	100%	100%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	114,5	97%	97%	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) Seizures (Ballif (2012) Hum Genet 131, 145) Epileptic encephalopathy (Mefford (2011) Ann Neurol 70, 974) Intellectual disability & seizures (Thierry (2012) Am J Med Genet A 158A, 1633) Thin corpus callosum, psychomotor delay & seizures (Selmer (2012) Eur J Med Genet 55,715)
HSD17B10	58,4	93%	86%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation,X-linked syndromic 10,300220
HSD17B4	97,8	96%	95%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1,233400
IDH2	122,9	100%	96%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	69,2	100%	94%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	138,2	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1,182250
IQSEC2	39,6	84%	71%	Mental retardation, X-linked 1, 309530
KANSL1	64,2	93%	84%	Koolen-De Vries syndrome, 610443
KCNA1	118,1	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	222,8	100%	100%	Epileptic encephalopathy, early infantile, 32,616366
KCNB1	164,4	99%	99%	Epileptic encephalopathy, early infantile, 26,616056
KCNC1	150,9	100%	100%	Epilepsy, progressive myoclonic 7,616187
KCNH1	116,8	100%	100%	Temple-Baraitser syndrome,611816 Zimmermann-Laband syndrome 1,135500

KCNJ10	170	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct,digenic,600791
KCNJ11	143	100%	100%	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582
KCNMA1	94	99%	96%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	77,6	98%	95%	Epileptic encephalopathy,early infantile,7,613720 Myokymia,121200 Seizures, benign neonatal, 1, 121200
KCNQ3	105	100%	99%	Seizures, benign neonatal, type 2, 121201
KCNT1	81,3	94%	91%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy,nocturnal frontal lobe,5,615005
KCTD7	92,7	72%	69%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	61,3	100%	95%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KPTN	96,2	100%	100%	Mental retardation, autosomal recessive 41, 615637
LGI1	149,9	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	115,8	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
MBD5	154,1	100%	100%	Mental retardation, autosomal dominant 1, 156200
MECP2	87	97%	86%	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	67,4	94%	86%	Lujan-Fryns syndrome,309520 Ohdo syndrome,X-linked,300895 Opitz-Kaveggia syndrome, 305450
MEF2C	106,8	100%	99%	Chromosome 5q14.3 deletion syndrome,613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	129,3	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement,616170
MOCS1	88,9	98%	93%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	117,7	99%	99%	Molybdenum cofactor deficiency, type B, 252150

MPDU1	125,7	100%	99%	Congenital disorder of glycosylation, type If
MTHFR	107,8	100%	99%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects,susceptibility to},601634 {Schizophrenia,susceptibility to},181500 {Thromboembolism,susceptibility to},188050
MTOR	108,1	100%	99%	No OMIM phenotype Lennox-Gastaut syndrome (Allen(2013) Nature 501, 217)
NDUFA1	102,1	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	103,8	93%	87%	Mitochondrial complex I deficiency, 252010
NDUFAF1	121,1	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	54,1	100%	95%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	141,4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	71,9	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFAF5	131,4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFB3	0,5	0%	0%	Mitochondrial complex I deficiency, 252010
NDUFB9	115,9	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS1	83,1	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	136,9	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS3	150,2	96%	91%	Mitochondrial complex I deficiency, 252010
NDUFS4	134,4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS6	127,1	96%	93%	Mitochondrial complex I deficiency, 252010
NDUFV1	68,9	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	116,4	98%	98%	Mitochondrial complex I deficiency, 252010
NECAP1	115,4	100%	100%	?Epileptic encephalopathy, early infantile,21, 615833
NEDD4L	117,5	100%	100%	No OMIM phenotype
NGLY1	117,3	100%	98%	Congenital disorder of glycosylation, type Iv
NHLRC1	127	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NRXN1	130,2	99%	98%	Pitt-Hopkins-like syndrome 2, 614325
NUBPL	96,3	100%	99%	Mitochondrial complex I deficiency, 252010
OFD1	39,7	87%	78%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome,type 2,300209

OPHN1	58	98%	90%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	56,2	100%	99%	Mental retardation, X-linked 30/47, 300558
PC	102,3	96%	91%	Pyruvate carboxylase deficiency, 266150
PCDH19	79,3	99%	98%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	68	95%	91%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	117,9	100%	97%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	174,3	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	51,3	100%	97%	MODY,type IV,606392 Pancreatic agenesis 1,260370 {Diabetes mellitus,type II,susceptibility to},125853
PET100	83,3	100%	99%	Mitochondrial complex IV deficiency, 220110
PEX1	127	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD),601539
PEX10	82,5	97%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B,614871
PEX12	137,3	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B,266510
PEX13	153,2	96%	93%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B,614885
PEX14	99,9	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100,1	89%	86%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B,614877
PEX19	113,6	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	123,1	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B,614873
PEX3	144,5	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	95,1	98%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B,202370
PEX6	97,5	98%	88%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B,614863
PGAP3	71,3	95%	88%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	80,6	100%	90%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	89,3	100%	100%	Neu-Laxova syndrome 1,256520 Phosphoglycerate dehydrogenase deficiency, 601815

PIGA	78,7	99%	97%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	111,3	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	115,5	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	134,8	100%	100%	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PLA2G6	84	99%	91%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B,610217 Parkinson disease 14,autosomal recessive,612953
PLCB1	121	100%	94%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	42	96%	84%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2,X-linked,312920
PMM2	101	100%	100%	Congenital disorder of glycosylation, type Ia
PNKP	81,2	100%	99%	Ataxia-oculomotor apraxia,616267 Microcephaly, seizures and developmental delay, 613402
PNPO	73,3	100%	99%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	94,1	98%	95%	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,157640 Progressive external ophthalmoplegia, autosomal recessive, 258450
PPP2R1A	98,1	93%	92%	Mental retardation, autosomal dominant 36,616362
PPT1	77,4	100%	99%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	70,1	98%	96%	Renpenning syndrome, 309500
PRICKLE1	114,1	100%	99%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	120,2	95%	94%	Epilepsy, progressive myoclonic 5,613832
PRRT2	91,4	100%	100%	Convulsions,familial infantile,with paroxysmal choreoathetosis,602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures,benign familial infantile, 2,605751
PURA	118,1	100%	97%	Mental retardation, autosomal dominant 31
QARS	128	100%	100%	Microcephaly, progressive,seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	105,7	100%	100%	?Waisman syndrome,311510 Mental retardation, X-linked 72, 300271
RARS2	91,4	100%	100%	Pontocerebellar hypoplasia, type 6, 611523

RNASEH2A	98,2	100%	93%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	106,4	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	161,7	100%	100%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	98,5	95%	95%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	52,3	100%	92%	Coffin-Lowry syndrome, 303600 Mental retardation,X-linked 19,300844
RRM2B	130,9	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type),612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal dominant, 5,613077
RYR3	111,2	99%	99%	No OMIM phenotype Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360) Hyperinsulinism (Proverbio (2013) PLoS One 8, e68740) Schizophrenia (Fromer (2014) Nature 506, 179) Lennox-Gastaut syndrome (Appenzeller (2014) Am J Hum Genet 95, 360)
SAMHD1	122,5	100%	99%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2,614415
SCARB2	100,5	100%	97%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	120,6	99%	98%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures,familial,3A,604403 Migraine,familial hemiplegic,3,609634
SCN1B	121,8	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	129,6	100%	99%	Epileptic encephalopathy,early infantile,11,613721 Seizures, benign familial infantile, 3, 607745
SCN8A	146,9	100%	99%	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy,early infantile,13,614558

SCN9A	115,3	100%	99%	Epilepsy,generalized,with febrile seizures plus,type 7,613863 Erythermalgia, primary, 133020 Febrile seizures,familial,3B,613863 HSAN2D,autosomal recessive,243000 Insensitivity to pain, congenital,243000 Paroxysmal extreme pain disorder,167400 Small fiber neuropathy,133020 {Dravet syndrome,modifier of},607208
SIK1	62,9	98%	88%	Epileptic encephalopathy, early infantile, 30,616341
SLC13A5	87,8	100%	99%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	158,8	100%	99%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia,familial,7,610021 Monocarboxylate transporter 1 deficiency,616095
SLC19A3	113,8	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	79,4	89%	83%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	111,4	94%	84%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	85	100%	96%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	96	100%	100%	Dystonia 9,601042 GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized,susceptibility,12}
SLC35A2	53,5	100%	96%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	100,9	99%	98%	Myoclonic-atonic epilepsy,616421
SLC6A8	5,4	10%	7%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	62,1	96%	92%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMC1A	72,3	96%	91%	Cornelia de Lange syndrome 2, 300590
SMS	17	66%	28%	Smith-Magenis syndrome, 182290
SPTAN1	104,7	100%	98%	Epileptic encephalopathy, early infantile, 5, 613477
SRPX2	42,7	93%	80%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	120,4	100%	100%	Epileptic encephalopathy,early infantile,15,615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	123,5	94%	91%	Amish infantile epilepsy syndrome, 609056
STXBP1	97,9	100%	99%	Epileptic encephalopathy, early infantile, 4, 612164

SUOX	197,1	100%	100%	Sulfite oxidase deficiency, 272300
SYN1	38,7	83%	62%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	59,3	93%	81%	Mental retardation, autosomal dominant 5, 612621
SYP	51	99%	88%	Mental retardation, X-linked 96, 300802
SZT2	110,5	99%	95%	Epileptic encephalopathy, early infantile, 18, 615476
TBC1D24	124,6	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	125,3	100%	98%	Hypoparathyroidism-retardation-dysmorphism syndrome,241410 Kenny-Caffey syndrome-1, 244460
TCF4	99,1	97%	97%	Corneal dystrophy,Fuchs endothelial 3,613267 Pitt-Hopkins syndrome, 610954
TDP2	126,9	100%	98%	No OMIM phenotype
TPP1	145,4	100%	97%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia,autosomal recessive 7,609270
TREX1	147,2	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	129	100%	98%	Hypomagnesemia 1, intestinal, 602014
TSC1	103,6	99%	98%	Focal cortical dysplasia, Taylor balloon cell type, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TUBB2A	46,3	100%	92%	Cortical dysplasia, complex, with other brain malformations 5, 615763
UBE3A	106,3	100%	100%	Angelman syndrome, 105830
WWOX	107	97%	97%	Epileptic encephalopathy, early infantile, 28,616211 Esophageal squamous cell carcinoma, somatic,133239 Spinocerebellar ataxia,autosomal recessive 12,614322
ZEB2	160,3	100%	100%	Mowat-Wilson syndrome, 235730

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : June 30th, 2015
This list is accurate for all panel versions starting with DG 2.3. (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