Epilepsie genpanel v1 (252 genen)

Gene	% covered > 30x	Associated phenotype description and OMIM ID
AARS	99,9	Epileptic encephalopathy, early infantile, 29,616339 Charcot-Marie-Tooth disease, axonal, type 2N,613287
ABAT	100,0	GABA-transaminase deficiency, 613163
ABCC8	100,0	Hyperinsulinemic hypoglycemia, familial, 1, 256450
ACY1	99,7	Aminoacylase 1 deficiency, 609924
ADSL	100,0	Adenylosuccinase deficiency, 103050
ALDH7A1	99,9	Epilepsy, pyridoxine-dependent, 266100
ALG1	99,8	Congenital disorder of glycosylation, type Ik,608540
ALG11	99,6	Congenital disorder of glycosylation, type Ip,613661
ALG13	67,7	Congenital disorder of glycosylation, type Is, 300884
ALG3	94,5	Congenital disorder of glycosylation, type Id,601110
ALG6	100,0	Congenital disorder of glycosylation, type Ic,603147
AMACR	99,8	Alpha-methylacyl-CoA racemase deficiency, 614307
AMT	76,6	Glycine encephalopathy, 605899
APOPT1	58,8	Mitochondrial complex IV deficiency, 220110
ARHGEF9	100,0	Epileptic encephalopathy, early infantile, 8, 300607
ARX	90,2	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	100,0	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy,159950
ATP1A2	99,7	Migraine, familial hemiplegic, 2, 602481 Migraine,familial basilar,602481 Alternating hemiplegia of childhood,104290
ATP1A3	100,0	Alternating hemiplegia of childhood 2,614820 CAPOS syndrome,601338 Dystonia-12,128235
ATP6AP2	99,7	?Mental retardation, X-linked, syndromic,Hedera type, 300423 ?Parkinsonism with spasticity,X-linked,300911
ATP7A	58,0	Menkes disease, 309400 Occipal horn syndrome,304150 Spinal muscular atrophy,distal,X-linked 3,300489
ATRX	99,7	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome,somatic,300448 Mental retardation-hypotonic facies syndrome,X-linked,309580
AUTS2	99,6	Mental retardation, autosomal dominant 26,615834
BOLA3	83,0	Multiple mitochondrial dysfunctions syndrome 2, 614299
BTD	100,0	Biotinidase deficiency, 253260
CACNA1A	100,0	Episodic ataxia,type 2,108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic,1,with progressive cerebellar ataxia,141500 Spinocerebellar ataxia 6,183086
CACNA2D2	94,6	No OMIM phenotype Epileptic encephalopathy (Pippucci, PLoS One. 2013 Dec 16;8(12):e82154)
CASK	100,0	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation,with or without nystagmus,300422 FG syndrome 4,300422
CDKL5	100,0	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	100,0	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	100,0	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	36,7	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction,susceptibility to},188890
CHRNB2	99,5	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	100,0	Hypomagnesemia 3, renal, 248250
CLDN19	100,0	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	99,7	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	99,8	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	96,4	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis,neuronal,Kufs type,adult onset,204300
CLN8	100,0	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8,Northern epilepsy variant,610003
CNNM2	100,0	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	74,7	?Epilepsy, familial adult myoclonic, 5, 615400
CNTNAP2	99,9	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1,610042 {Autism susceptibility 15},612100
COL4A3BP	99,2	Mental retardation, autosomal dominant 34,616351
COQ2	100,0	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to},146500

Gene	% covered > 30x	Associated phenotype description and OMIM ID
CPA6	100,0	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures,familial,11,614418
CPS1	99,6	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension,neonatal,susceptibility to},615371 {Venoocclusive disease after bone marrow transplantation}
CPT2	100,0	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	100,0	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	99,9	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	87,2	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	99,4	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	67,6	D-2-hydroxyglutaric aciduria, 600721
DCX	99,9	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DEPDC5	91,0	Epilepsy, familial focal, with variable foci, 604364
DLAT	100,0	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	100,0	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	99,9	Epileptic encephalopathy,early infantile,31,616346
DOCK7	98,9	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	100,0	Congenital disorder of glycosylation, type Ij,608093 Myasthenic syndrome,congenital,13,with tubular aggregates,614750
DPM1	100,0	Congenital disorder of glycosylation, type Ie,608799
DPM2	100,0	Congenital disorder of glycosylation, type Iu,615042
DPYD	99,8	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity,274270
DYNC1H1	100,0	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	99,8	Mental retardation, autosomal dominant 7, 614104
EEF1A2	99,9	Epileptic encephalopathy,early infantile,33,616409 Mental retardation,autosomal dominant 38,616393
EGF	99,6	Hypomagnesemia 4, renal, 611718
EHMT1	98,8	Kleefstra syndrome, 610253
EPM2A	98,0	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	100,0	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100,0	Combined oxidative phosphorylation deficiency 14, 614946
FASN	100,0	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	98,9	Aarskog-Scott syndrome, 305400 Mental retardation,X-linked syndromic 16,305400
FLNA	99,8	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	99,8	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	99,6	Rett syndrome, congenital variant, 613454
FOXRED1	83,6	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency,252010
FXYD2	100,0	Hypomagnesemia-2, renal, 154020
GABBR2	95,8	{Nicotine dependance},188890 Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360)
GABRA1	100,0	Epileptic encephalopathy,early infantile,19,615744 {Epilepsy,childhood absense,susceptibility to,4},611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	60,3	{Epilepsy,childhood absence, susceptibility to, 5},612269 Epileptic encephalopathy (Epi4K consortium, Nature. 2013 Sep 12;501(7466):217-21)
GABRG2	91,9	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures,familial,8,611277 {Epilepsy,childhood absence,susceptibility to,2},607681
GAMT	99,1	Cerebral creatine deficiency syndrome 2, 612736
GCK	99,8	Diabetes mellitus,noninsulin-dependent,late onset,125853 Diabetes mellitus,permanent neonatal,606176 Hyperinsulinemic hypoglycemia,familial,3,602485 MODY, type II, 125851
GCSH	41,4	Glycine encephalopathy, 605899
GLDC	99,9	Glycine encephalopathy, 605899

Gene	% covered > 30x	Associated phenotype description and OMIM ID
GLRA1	99,9	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	100,0	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	99,6	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	98,7	Epileptic encephalopathy, early infantile, 17, 615473
GOSR2	98,4	Epilepsy, progressive myoclonic 6
GPC3	100,0	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor,somatic,194070
GPHN	100,0	Molybdenum cofactor deficiency, type C, 252150
GRIA3	99,9	Mental retardation, X-linked 94, 300699
GRIN1	100,0	Mental retardation, autosomal dominant 8, 614254
GRIN2A	100,0	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	100,0	Mental retardation, autosomal dominant 6, 613970 Epileptic encephalopathy,early infantile,27,616139
GRN	100,0	Aphasia,primary progressive,607485 Ceroid lipofuscinosis,neuronal,11,614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HADH	98,2	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCN1	100,0	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	99,9	Brachydactyly-mental retardation syndrome, 600430
HLCS	99,3	Holocarboxylase synthetase deficiency, 253270
HNRNPU	100,0	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	99,7	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation,X-linked syndromic 10,300220
HSD17B4	99,9	D-bifunctional protein deficiency, 261515 Perrault syndrome 1,233400
IDH2	99,1	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	100,0	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100,0	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1,182250
IQSEC2	99,7	Mental retardation, X-linked 1, 309530
KANSL1	99,8	Koolen-De Vries syndrome, 610443
KCNA1	100,0	Episodic ataxia/myokymia syndrome, 160120
KCNA2	98,3	Epileptic encephalopathy, early infantile, 32,616366
KCNB1	100,0	Epileptic encephalopathy, early infantile, 26,616056
KCNC1	98,4	Epilepsy, progressive myoclonic 7,616187
KCNH1	95,8	Temple-Baraitser syndrome,611816 Zimmermann-Laband syndrome 1,135500
KCNJ10	100,0	SESAME syndrome, 612780 Enlarged vestibular aqueduct,digenic,600791
KCNJ11	100,0	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	100,0	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	100,0	Epileptic encephalopathy,early infantile,7,613720 Myokymia,121200 Seizures, benign neonatal, 1, 121200
KCNQ3	99,1	Seizures, benign neonatal, type 2, 121201
KCNT1	99,2	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy,nocturnal frontal lobe,5,615005
KCTD7	99,9	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	100,0	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KPTN	84,5	Mental retardation, autosomal recessive 41, 615637
LGI1	79,2	Epilepsy, familial temporal lobe, 1, 600512
LIAS	99,7	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
MBD5	100,0	Mental retardation, autosomal dominant 1, 156200
MECP2	100,0	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	99,9	Lujan-Fryns syndrome,309520 Ohdo syndrome,X-linked,300895 Opitz-Kaveggia syndrome, 305450
MEF2C	99,9	Chromosome 5q14.3 deletion syndrome,613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	100,0	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement,616170

Gene	% covered > 30x	Associated phenotype description and OMIM ID
MOCS1	55,9	Molybdenum cofactor deficiency, type A, 252150
MOCS2	100,0	Molybdenum cofactor deficiency, type B, 252150
MPDU1	85,2	Congenital disorder of glycosylation, type If
MTHFR	99,9	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects,susceptibility to},601634 {Schizophrenia,susceptibility to},181500 {Thromboembolism,susceptibility to},188050
MTOR	94,4	No OMIM phenotype Lennox-Gastaut syndrome (Allen(2013) Nature 501, 217)
NDUFA1	99,4	Mitochondrial complex I deficiency, 252010
NDUFA11	72,4	Mitochondrial complex I deficiency, 252010
NDUFAF1	70,6	Mitochondrial complex I deficiency, 252010
NDUFAF2	100,0	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	100,0	Mitochondrial complex I deficiency, 252010
NDUFAF4	99,7	Mitochondrial complex I deficiency, 252010
NDUFAF5	55,5	Mitochondrial complex I deficiency, 252010
NDUFB3	100,0	Mitochondrial complex I deficiency, 252010
NDUFB9	100,0	Mitochondrial complex I deficiency, 252010
NDUFS1	99,9	Mitochondrial complex I deficiency, 252010
NDUFS2	99,8	Mitochondrial complex I deficiency, 252010
NDUFS3	100,0	Mitochondrial complex I deficiency, 252010
NDUFS4	100,0	Mitochondrial complex I deficiency, 252010
NDUFS6	99,8	Mitochondrial complex I deficiency, 252010
NDUFV1	99,9	Mitochondrial complex I deficiency, 252010
NDUFV2	100,0	Mitochondrial complex I deficiency, 252010
NECAP1	44,3	?Epileptic encephalopathy, early infantile,21, 615833
NEDD4L	100,0	No OMIM phenotype
NGLY1	99,3	Congenital disorder of glycosylation, type Iv
NHLRC1	100,0	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NRXN1	99,2	Pitt-Hopkins-like syndrome 2, 614325
NUBPL	46,4	Mitochondrial complex I deficiency, 252010
OFD1	100,0	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome,type 2,300209
OPHN1	99,9	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	100,0	Mental retardation, X-linked 30/47, 300558
PC	99,3	Pyruvate carboxylase deficiency, 266150
PCDH19	99,8	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	99,9	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	80,5	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	100,0	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	98,9	MODY,type IV,606392 Pancreatic agenesis 1,260370 {Diabetes mellitus,type II,susceptibility to},125853
PET100	84,2	Mitochondrial complex IV deficiency, 220110
PEX1	100,0	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD),601539
PEX10	98,8	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B,614871
PEX12	99,9	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B,266510
PEX13	100,0	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B,614885
PEX14	99,9	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100,0	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B,614877
PEX19	31,9	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	100,0	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B,614873
PEX3	100,0	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	100,0	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B,202370
PEX6	100,0	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B,614863
PGAP3	80,0	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	100,0	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	99,5	Neu-Laxova syndrome 1,256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	48,1	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	100,0	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100,0	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	84,9	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399

Gene	% covered > 30x	Associated phenotype description and OMIM ID
PLA2G6	100,0	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B,610217 Parkinson disease 14,autosomal recessive,612953
PLCB1	100,0	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	99,7	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2,X-linked,312920
PMM2	100,0	Congenital disorder of glycosylation, type Ia
PNKP	100,0	Ataxia-oculomotor apraxia,616267 Microcephaly, seizures and developmental delay, 613402
PNPO	97,3	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	100,0	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	80,7	Mental retardation, autosomal dominant 36,616362
PPT1	99,3	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100,0	Renpenning syndrome, 309500
PRICKLE1	99,8	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	100,0	Epilepsy, progressive myoclonic 5,613832
PRRT2	99,7	Convulsions,familial infantile,with paroxysmal choreoathetosis,602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures,benign familial infantile, 2,605751
PURA	80,7	Mental retardation, autosomal dominant 31
QARS	83,2	Microcephaly, progressive,seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	100,0	?Waisman syndrome,311510 Mental retardation, X-linked 72, 300271
RARS2	100,0	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	100,0	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,0	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100,0	Aicardi-Goutieres syndrome 3, 610329
ROGDI	70,1	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	100,0	Coffin-Lowry syndrome, 303600 Mental retardation,X-linked 19,300844
RRM2B	99,9	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	99,9	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	99,9	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2,614415
SCARB2	100,0	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	100,0	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures,familial,3A,604403 Migraine,familial hemiplegic,3,609634
SCN1B	93,4	Atrial fibrillation,familial,13,615366 Brugada syndrome 5,612838 Cardiac conduction defect,nonspecific,612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	100,0	Epileptic encephalopathy,early infantile,11,613721 Seizures, benign familial infantile, 3, 607745
SCN8A	100,0	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy,early infantile,13,614558
SCN9A	99,9	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	100,0	Epileptic encephalopathy, early infantile, 30,616341
SLC13A5	99,9	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	99,7	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia,familial,7,610021 Monocarboxylate transporter 1 deficiency,616095
SLC19A3	99,9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	53,1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	99,9	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100,0	Epileptic encephalopathy, early infantile, 3, 609304

Gene	% covered > 30x	Associated phenotype description and OMIM ID
SLC2A1	97,4	Dystonia 9,601042 GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized,susceptibility,12}
SLC35A2	100,0	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	99,6	Myoclonic-atonic epilepsy,616421
SLC6A8	99,2	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	99,1	Mental retardation, X-linked syndromic, Christianson type, 300243
SMC1A	99,9	Cornelia de Lange syndrome 2, 300590
SMS	99,7	Smith-Magenis syndrome, 182290
SPTAN1	100,0	Epileptic encephalopathy, early infantile, 5, 613477
SRPX2	99,7	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	65,2	Epileptic encephalopathy,early infantile,15,615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	92,3	Amish infantile epilepsy syndrome, 609056
STXBP1	99,8	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	99,6	Sulfite oxidase deficiency, 272300
SYN1	98,7	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	100,0	Mental retardation, autosomal dominant 5, 612621
SYP	100,0	Mental retardation, X-linked 96, 300802
SZT2	87,2	Epileptic encephalopathy, early infantile, 18, 615476
TBC1D24	100,0	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	99,9	Hypoparathyroidism-retardation-dysmorphism syndrome,241410 Kenny-Caffey syndrome-1, 244460
TCF4	100,0	Corneal dystrophy,Fuchs endothelial 3,613267 Pitt-Hopkins syndrome, 610954
TDP2	100,0	No OMIM phenotype
TPP1	99,7	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia,autosomal recessive 7,609270
TREX1	100,0	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus,610448 Vasculopathy,retinal,with cerebral leukodystrophy,192315 {Systemic lupus erythematosus,susceptibility to},152700
TRPM6	100,0	Hypomagnesemia 1, intestinal, 602014
TSC1	99,9	Focal cortical dysplasia, Taylor balloon cell type, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TUBB2A	100,0	Cortical dysplasia, complex, with other brain malformations 5, 615763
UBE3A	100,0	Angelman syndrome, 105830
WWOX	79,6	Epileptic encephalopathy, early infantile, 28,616211 Esophageal squamous cell carcinoma, somatic,133239 Spinocerebellar ataxia,autosomal recessive 12,614322
ZEB2	62,6	Mowat-Wilson syndrome, 235730

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Genes in bold are core genes
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
"No OMIM phenotype" signifies a gene without a current OMIM association
OMIM phenotype descriptions between {} signify risk factors