Panelinformation Epilepsi in-silico

Version: Epilepsi. V2.0

Gen	Associerad Fenotyp, MIM# (OMIM)
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABAT	GABA-transaminase deficiency, 613163
ABCC8	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACY1	Aminoacylase 1 deficiency, 609924
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADCY5	Dyskinesia, familial, with facial myokymia, 606703
ADRA2B	Epilepsy, myoclonic, familial adult, 2, 607876
ADSL	Adenylosuccinase deficiency, 103050
AFG3L2	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGTR2	ANGIOTENSIN II RECEPTOR, TYPE 2; AGTR2, 300034 Ylisaukko-oja, T. Identification of two AGTR2 mutations in male patients with non-syndromic mental retardation. Hum. Genet. 114: 211-213, 2004.
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100
ALG1	Congenital disorder of glycosylation, type Ik, 608540
ALG11	Congenital disorder of glycosylation, type Ip, 613661
ALG13	Epileptic encephalopathy, early infantile, 36, 300884
ALG3	Congenital disorder of glycosylation, type Id, 601110
ALG6	Congenital disorder of glycosylation, type Ic, 603147
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMT	Glycine encephalopathy, 605899
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067
APOPT1	Mitochondrial complex IV deficiency, 220110
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607
ARID1B	Coffin-Siris syndrome 1, 135900
ARV1	Epileptic encephalopathy, early infantile, 38, 617020
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	Argininosuccinic aciduria, 207900
ASNS	Asparagine synthetase deficiency, 615574
ATP13A2	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP6AP2	?Parkinsonism with spasticity, X-linked, 300911 ?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	Mental retardation, autosomal dominant 26,615834
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRWD3	Mental retardation, X-linked 93, 300659
BSCL2	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BTD	Biotinidase deficiency, 253260
CACNA1A	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1E	No OMIM phenotype ?Epileptic encephalopathy with infantile spasms (Helbig (2016) Genet Med Epub,Epub) ?Autism (O'Roak (2012) Nature 485,246)
CACNA2D1	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, ALPHA-2/DELTA SUBUNIT 1; CACNA2D1, 114204 Vergult, S. Genomic aberrations of the CACNA2D1 gene in three patients with epilepsy and intellectual disability. Europ. J. Hum. Genet. 23: 628-632, 2015.
CACNA2D2	No OMIM phenotype Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154) ?Schizophrenia (Purcell (2014) Nature 506, 185)
CAD	Epileptic encephalopathy, early infantile, 50, 616457
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672
CERS1	?Epilepsy, progressive myoclonic, 8, 616230
CHD2	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNA7	Chromosome 15q13.3 microdeletion syndrome, 612001 Epilepsy, juvenile myoclonic 604827 {Epilepsy, idiopathic generalized, susceptibility to, 7}, 604827 {Schizophrenia, susceptibility to, 13}, 613025

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	Hypomagnesemia 3, renal, 248250
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNKSR2	CONNECTOR ENHANCER OF KSR 2; CNKSR2, 300724 - X-linked ID and seizures (recently described)
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	Mental retardation, autosomal dominant 34, 616351
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016
CPA6	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT2	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CRADD	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	D-2-hydroxyglutaric aciduria, 600721
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828
DENND5A	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	Epilepsy, familial focal, with variable foci, 604364
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	Epileptic encephalopathy, early infantile, 31, 616346

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DOCK7	Epileptic encephalopathy, early infantile, 23, 615859
DOLK	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	Congenital disorder of glycosylation, type Ie, 608799
DPM2	Congenital disorder of glycosylation, type Iu, 615042
DPYD	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYRK1A	Mental retardation, autosomal dominant 7, 614104
EEF1A2	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EGF	Hypomagnesemia 4, renal, 611718
EHMT1	Kleefstra syndrome, 610253
EIF2S3	Mental retardation, X-linked, syndromic, Borck type, 300987
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF12	Epileptic encephalopathy, early infantile, 47, 617166
FLNA	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	Rett syndrome, congenital variant, 613454
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRRS1L	Epileptic encephalopathy, early infantile, 37, 616981
FXYD2	Hypomagnesemia 2, renal, 154020
GABRA1	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB1	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	?Intellectual disability and epilepsy, 600232.0001 (Srivastava et al. 2014)
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRD	{Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to}, 613060 {Epilepsy, idiopathic generalized, 10}, 613060

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

	{Epilepsy, juvenile myoclonic, susceptibility to}, 613060
GABRG2	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAL	?Epilepsy, familial temporal lobe, 8, 616461
GAMT	Cerebral creatine deficiency syndrome 2, 612736
GATM	Cerebral creatine deficiency syndrome 3,612718
GCK	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCSH	Glycine encephalopathy, 605899
GLDC	Glycine encephalopathy, 605899
GLRA1	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473
GNB1	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB5	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GOSR2	Epilepsy, progressive myoclonic 6, 614018
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	Molybdenum cofactor deficiency C, 615501
GRIA3	Mental retardation, X-linked 94, 300699
GRIN1	Mental retardation, autosomal dominant 8, 614254
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	Epileptic encephalopathy, early infantile, 46, 617162
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GUF1	?Epileptic encephalopathy, early infantile, 40, 617065
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCN1	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	No OMIM phenotype Anorexia nervosa/bulimia nervosa (Cui (2013) J Clin Invest 123,4706) Brachydactyly mental retardation syndrome (Williams (2010) Am J Hum Genet 87, 219) ?Autism spectrum disorder (Pinto (2014) Am J Hum Genet 94, 677)
HECW2	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HLCS	Holocarboxylase synthetase deficiency, 253270
HNRNPH2	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	Mental retardation, X-linked 1/78, 309530
ITPA	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	Koolen-De Vries syndrome, 610443
KCNA1	Episodic ataxia/myokymia syndrome, 160120
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	Epilepsy, progressive myoclonic 7, 616187
KCNH1	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	Seizures, benign neonatal, type 2, 121201
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KIF1A	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF5A	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KPTN	Mental retardation, autosomal recessive 41, 615637
LGI1	Epilepsy, familial temporal lobe, 1, 600512
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462
LMNB2	?Epilepsy, progressive myoclonic, 9, 616540 {Lipodystrophy, partial, acquired, susceptibility to}, 608709
MAGEL2	Schaaf-Yang syndrome, 615547
MBD5	Mental retardation, autosomal dominant 1, 156200
MBOAT7	Mental retardation, autosomal recessive 57, 617188
MDH2	Epileptic encephalopathy, early infantile, 51, 617339

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

MECP2	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFF	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	Molybdenum cofactor deficiency A, 252150
MOCS2	Molybdenum cofactor deficiency B, 252160
MPDU1	Congenital disorder of glycosylation, type If, 609180
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTOR	Smith-Kingsmore syndrome, 616638
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAPB	Conroy, J. NAPB--a novel SNARE-associated protein for early-onset epileptic encephalopathy. Clin. Genet. 89: E1-E3, 2016.
NDUFA1	Mitochondrial complex I deficiency, 252010
NDUFA11	Mitochondrial complex I deficiency, 252010
NDUFAF1	Mitochondrial complex I deficiency, 252010
NDUFAF2	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	Mitochondrial complex I deficiency, 252010
NDUFAF4	Mitochondrial complex I deficiency, 252010
NDUFAF5	Mitochondrial complex 1 deficiency, 252010
NDUFB3	Mitochondrial complex I deficiency, 252010
NDUFB9	?Mitochondrial complex I deficiency, 252010
NDUFS1	Mitochondrial complex I deficiency, 252010
NDUFS2	Mitochondrial complex I deficiency, 252010
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	Mitochondrial complex I deficiency, 252010
NDUFV1	Mitochondrial complex I deficiency, 252010
NDUFV2	Mitochondrial complex I deficiency, 252010
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	Periventricular nodular heterotopia 7, 617201

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

NEXMIF	Mental retardation, X-linked 98, 300912
NGLY1	Congenital disorder of deglycosylation, 615273
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	Epilepsy, familial focal, with variable foci, 3, 617118
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 Cardoso, C. Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. Neurology 72: 784-792, 2009.
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	Mitochondrial complex I deficiency, 252010
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OTUD6B	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAK3	Mental retardation, X-linked 30/47, 300558
PC	Pyruvate carboxylase deficiency, 266150
PCDH12	Microcephaly, seizures, spasticity, and brain calcification, 605622
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PET100	Mitochondrial complex IV deficiency, 220110
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGG	Mental retardation, autosomal recessive 53, 616917
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGQ	Martin, HC. Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. Hum. Molec. Genet. 23: 3200-3211, 2014.
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	?Hyperphosphatasia with mental retardation syndrome 5, 616025
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	Congenital disorder of glycosylation, type Ia, 212065
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	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, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PPP2R1A	Mental retardation, autosomal dominant 36, 616362
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	Renpenning syndrome, 309500
PRDM8	?Epilepsy, progressive myoclonic, 10, 616640
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	No OMIM phenotype ?Autism spectrum disorder (Sowers (2013) Mol Psychiatry 18, 1077) ?Myoclonus epilepsy (Tao (2011) Am J Hum Genet 88,138)
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRUNE1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PURA	Mental retardation, autosomal dominant 31, 616158
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

RAB39B	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RARS2	Pontocerebellar hypoplasia, type 6, 611523
RBSN	Single point mutation in rabenosyn-5 in a female with intractable seizures and evidence of defective endocytotic trafficking (Stockler et al. 2014).
RELN	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
RFT1	Congenital disorder of glycosylation, type In (including epilepsy), 612015
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329
ROGDI	Kohlschutter-Tonz syndrome, 226750
RORB	RAR-RELATED ORPHAN RECEPTOR B; RORB, 601972 (Epilepsy familial ± ID - recently described)
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	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
RUSC2	RUN DOMAIN AND SH3 DOMAIN-CONTAINING PROTEIN 2; RUSC2, 611053 Mental retard AR 61
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	SODIUM CHANNEL, VOLTAGE-GATED, TYPE III, ALPHA SUBUNIT; SCN3A, 182391 Epil encephalop
SCN8A	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SCN9A	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
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies (including epilepsy), 604218 Neuroserpin mutation S52R causes neuroserpin accumulation in neurons and is associated with progressive myoclonus epilepsy. Takao et al. 2000

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15} 613950
SIK1	Epileptic encephalopathy, early infantile, 30, 616341
SLC12A5	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC13A5	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721
SLC6A1	Myoclonic-atonic epilepsy, 616421
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	Nicolaides-Baraitser syndrome, 601358
SMC1A	Cornelia de Lange syndrome 2,300590
SMS	Mental retardation, X-linked, Snyder-Robinson type, 309583
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	Amish infantile epilepsy syndrome, 609056
STAMBP	Microcephaly-capillary malformation syndrome (including early-onset refractory epilepsy), 614261
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STX1B	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164
STYXL1	SERINE/THREONINE/TYROSINE-INTERACTING PROTEIN-LIKE 1; STYXL1, 616695 ID, epilepsy
SUMF1	Multiple sulfatase deficiency, 272200
SUOX	Sulfite oxidase deficiency, 272300
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	Mental retardation, autosomal dominant 5, 612621
SYNJ1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset 615530
SYP	Mental retardation, X-linked 96, 300802
SZT2	Epileptic encephalopathy, early infantile, 18, 615476

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00

Panelinformation Epilepsi in-silico

TANGO2	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TBC1D24	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOOR syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	Spinocerebellar ataxia, autosomal recessive, 616949
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM8	TRIPARTITE MOTIF-CONTAINING PROTEIN 8; TRIM8, 606125 - Early onset epileptic encephal.(recent)
TRPM6	Hypomagnesemia 1, intestinal, 602014
TSC1	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	Lissencephaly 3, 611603
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44 617132
UBE3A	Angelman syndrome, 105830
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
WDR45	Neurodegeneration with brain iron accumulation 5, 300894
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
ZBTB18	Mental retardation, autosomal dominant 22, 612337
ZEB2	Mowat-Wilson syndrome, 235730
ZNHIT3	PEHO syndrome, 260565

www.sahlgrenska.se

VÄSTRA GÖTALANDSREGIONEN
SAHLGRENSKA UNIVERSITETSSJUKHUSET

Klinisk genetik och genomik
Sahlgrenska Universitetssjukhuset
413 45 Göteborg

TELEFON växel 031-342 00 00