PANEL Epilepsie zonder ontwikkelingsachterstand, familiale epilepsie (FE) v1

Gen	NCBI gen ID	Overerving	OMIM
ADRA2B	151	AD	Epilepsy, myoclonic, familial adult, 2, 607876
ATP1A2	477	AD	Alternating hemiplegia of childhood 1, 104290|Migraine familial basilar, 602481|Migraine familial hemiplegic 2, 602481
ATP1A3	478	AD	Alternating hemiplegia of childhood 2, 614820 | CAPOS syndrome, 601338 | Dystonia-12, 128235
CHRNA2	1135	AD	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	1137	AD	Epilepsy, nocturnal frontal lobe, 1 600513 | Nicotine addiction, susceptibility to, 188890
CHRNB2	1141	AD	Epilepsy, nocturnal frontal lobe, 3, 605375
DEPDC5	9681	AD	Epilepsy, familial focal, with variable foci 1, 604364
FLNA	2316	XD/XR	Cardiac valvular dysplasia, 314400|Frontometaphyseal dysplasia, 305620 | Heterotopia, periventricular, 300049 | Melnick-Needles syndrome, 309350
GABRA1	2554	AD	Epileptic encephalopathy, early infantile, 19, 615744 |Epilepsy, juvenile myoclonic, susceptibility to 5, 611136 | Epilepsy, childhood absence, susceptibility to, 4, 611136
GABRB3	2562	AD	{Epilepsy, childhood absence, susceptibility to, 5}, 612269|Epileptic encephalopathy, early infantile, 43, 617113
GABRG2	2566	AD	Febrile seizures, familial, 8, 611277 | Epilepsy, generalized, with febrile seizures plus, type 3, 611277 |Epilepsy, childhood absence, susceptibility to, 2, 607681
GRIN2A	2903	AD	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
HCN1	348980	AD	Epileptic encephalopathy, early infantile, 24, 615871
KCNMA1	3778	AD/AR	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 | Cerebellar atrophy, developmental delay, and seizures, 617643 | Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 | Liang-Wang syndrome, 618729
KCNQ2	3785	AD	Epileptic encephalopathy, early infantile, 7, 613720| Seizures, benign neonatal, 1, 121200| Myokymia, 121200
KCNQ3	3786	AD	Seizures, benign neonatal, type 2, 121201
KCNT1	57582	AD	Epilepsy, nocturnal frontal lobe, 5, 615005|Epileptic encephalopathy, early infantile, 14, 614959
LGI1	9211	AD	Epilepsy, familial temporal lobe, 1, 600512
MTOR	2475	AD	Smith-Kingsmore syndrome 616638 | Focal cortical dysplasia, type II, somatic, 607341
NEXMIF	340533	X-linked	Mental retardation, X-linked 98, 300912
NPRL2	10641	AD	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	8131	AD	Epilepsy, familial focal, with variable foci 3, 617118
PCDH19	57526	X-linked	Epileptic encephalopathy, early infantile, 9, 300088
PRRT2	112476	AD	Seizures, benign familial infantile, 2, 605751| Episodic kinesigenic dyskinesia 1, 128200 |Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
RELN	5649	AD/AR	AR Lissencephaly 2 (Norman-Roberts type), 257320 | AD {Epilepsy, familial temporal lobe, 7}, 616436
RORB	6096	AD	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357

SCN1A	6323	AD	Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208| Epilepsy, generalized, with febrile seizures plus, type 2, 604403 | Febrile seizures, familial, 3A, 604403 |Migraine, familial hemiplegic, 3, 609634
SCN1B	6324	AD/AR	AD Epilepsy, generalized, with febrile seizures plus, type 1, 604233 | AR Epileptic encephalopathy, early infantile, 52, 617350 | Epileptic encephalopathy, early infantile, 52, 617350
SCN2A	6326	AD	Epileptic encephalopathy, early infantile, 11, 613721| Seizures, benign familial infantile, 3, 607745
SCN3A	6328	AD	Epilepsy, familial focal, with variable foci 4, 617935 | Epileptic encephalopathy, early infantile, 62, 617938
SCN8A	6334	AD	Seizures, benign familial infantile, 5, 617080 |Epileptic encephalopathy, early infantile, 13, 614558| Cognitive impairment with or without cerebellar ataxia, 614306
SLC2A1	6513	AD	{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 | Dystonia 9, 601042 | GLUT1 deficiency syndrome 2, childhood onset, 612126 | GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 | Stomatin-deficient cryohydrocytosis with neurologic defects, 608885
STX1B	112755	AD	Generalized epilepsy with febrile seizures plus, type 9, 616172
TBC1D24	57465	AR	Myoclonic epilepsy, infantile, familial, 605021 | Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 | Epileptic encephalopathy, early infantile, 16, 615338 |DOORS syndrome, 220500
TSC1	7248	AD	Focal cortical dysplasia, type II, somatic, 607341 | Tuberous sclerosis-1, 191100 | Lymphangioleiomyomatosis, 606690
TSC2	7249	AD	?Focal cortical dysplasia, type II, somatic, 607341 |Tuberous sclerosis-2, 613254| Lymphangioleiomyomatosis, somatic, 606690