Paroxysmal Episodic Disorders panel

versie	V2 (53 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ADCY5	600293	Dyskinesia, familial, with facial myokymia, 606703 (3), Autosomal dominant
ATAD1	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
ATP1A2	182340	Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant
ATP1A3	182350	CAPOS syndrome, 601338 (3), Autosomal dominant; Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
BCKDHA	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
CACNA1A	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNB4	601949	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; Episodic ataxia, type 5, 613855 (3), Autosomal dominant
CEP290	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive
CHRNA4	118504	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
CHRNB2	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CNTNAP2	604569	{Autism susceptibility 15}, 612100 (3); Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; Cortical dysplasia-focal epilepsy syndrome, 610042 (3), Autosomal recessive

COL4A1	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant
CRH	122560	No OMIM phenotype
CSNK1D	600864	Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant
DBT	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
DEPDC5	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
DLAT	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DNMT1	126375	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant
ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
FGF14	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
GCH1	600225	Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive; Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal dominant, Autosomal recessive
GLRA1	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
GLRB	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GNAO1	139311	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
KCNA1	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNK18	613655	{Migraine, with or without aura, susceptibility to, 13}, 613656 (3), Autosomal dominant
KCNMA1	600150	Liang-Wang syndrome, 618729 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
KCNQ2	602235	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant

KCNT1	608167	Epilepsy, nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 14, 614959 (3), Autosomal dominant
NOTCH3	600276	?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant; Lateral meningocele syndrome, 130720 (3), Autosomal dominant
PDE10A	610652	Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive; Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHX	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PNKD	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
PRKN	602544	Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3); Adenocarcinoma of lung, somatic, 211980 (3)
PRRT2	614386	Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant
SACS	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
SCN1A	182389	Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 (3), Autosomal dominant
SCN2A	182390	Episodic ataxia, type 9, 618924 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 11, 613721 (3), Autosomal dominant; Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant
SCN4A	603967	Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant; Paramyotonia congenita, 168300 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant
SCN8A	600702	Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; ?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant

SETX	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SLC16A2	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC1A3	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
SLC20A2	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
SLC2A1	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant
SLC6A5	604159	Hyperekplexia 3, 614618 (3), Autosomal dominant, Autosomal recessive
TBC1D24	613577	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 16, 615338 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness , autosomal recessive 86, 614617 (3), Autosomal recessive
TREX1	606609	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; Chilblain lupus, 610448 (3), Autosomal dominant
UBR4	609890	No OMIM phenotype
VAMP2	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Aug 20, 2020

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.