

Paroxysmal Episodic Disorders panel
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versie	16-Oct-2018 (41 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ADCY5</i>	600293	Dyskinesia, familial, with facial myokymia, 606703 (3), Autosomal dominant
<i>ATAD1</i>	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
<i>ATP1A2</i>	182340	Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
<i>ATP1A3</i>	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
<i>BCKDHA</i>	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
<i>BCKDHB</i>	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
<i>CACNA1A</i>	601011	Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant
<i>CACNB4</i>	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
<i>CHRNA4</i>	118504	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
<i>CHRN2</i>	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
<i>COL4A1</i>	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 607595 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 1, 175780 (3), Autosomal dominant; ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Schizencephaly, 269160 (3)
<i>CSNK1D</i>	600864	Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant
<i>DBT</i>	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
<i>DEPDC5</i>	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant

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<i>DLAT</i>	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
<i>DLD</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>ECHS1</i>	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
<i>FGF14</i>	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
<i>GCH1</i>	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
<i>GLRA1</i>	138491	Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant
<i>GLRB</i>	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
<i>GNAO1</i>	139311	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
<i>KCNA1</i>	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
<i>KCNK18</i>	613655	{Migraine, with or without aura, susceptibility to, 13}, 613656 (3)
<i>KCNMA1</i>	600150	?Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
<i>KCNQ2</i>	602235	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant
<i>NOTCH3</i>	600276	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant; Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant
<i>PDHA1</i>	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
<i>PDHX</i>	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
<i>PNKD</i>	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
<i>PRKN (PARK2)</i>	602544	Adenocarcinoma of lung, somatic, 211980 (3); Adenocarcinoma, ovarian, somatic, 167000 (3); {Leprosy, susceptibility to}, 607572 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive
<i>PRRT2</i>	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant

<i>SCN1A</i>	182389	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant
<i>SCN2A</i>	182390	Epileptic encephalopathy, early infantile, 11, 613721 (3), Autosomal dominant; Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant
<i>SCN8A</i>	600702	?Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant
<i>SLC16A2</i>	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<i>SLC1A3</i>	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
<i>SLC2A1</i>	138140	Dystonia 9, 601042 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant
<i>SLC6A5</i>	604159	Hyperekplexia 3, 614618 (3), Autosomal recessive, Autosomal dominant
<i>TREX1</i>	606609	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
<i>UBR4</i>	609890	No OMIM phenotype

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: July 04, 2018

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.