Paroxysmal Episodic Disorders

Gene panel

Gene panel information

Gene panel	**Paroxysmal Episodic Disorders**
Version	3
Total genes	63
Activation date	Thursday 04 april 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
ADCY5	99.98 %	600293	Dyskinesia with orofacial involvement, autosomal dominant, 606703 (3), Autosomal dominant; Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 (3), Autosomal recessive; Dyskinesia with orofacial involvement, autosomal recessive, 619647 (3), Autosomal recessive
ALPK1	99.92 %	607347	ROSAH syndrome, 614979 (3), Autosomal dominant
ANO3	99.98 %	610110	Dystonia 24, 615034 (3), Autosomal dominant
ATAD1	99.85 %	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
ATP1A2	99.85 %	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	99.98 %	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant
ATP7B	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
BCKDHA	99.97 %	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	99.73 %	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
CACNA1A	98.16 %	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNB4	99.2 %	601949	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; Episodic ataxia, type 5, 613855 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant
CEP290	98.1 %	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CHRNA4	100 %	118504	{Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant
CHRNB2	99.99 %	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)

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CLCN2	100 %	600570	Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant
CNTNAP2	99.99 %	604569	Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3)
COL4A1	99.99 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
CSNK1D	99.99 %	600864	Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant
DARS2	98.31 %	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DBT	94.51 %	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
DEPDC5	99.18 %	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
DLAT	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DNMT1	99.13 %	126375	Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant; Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant
ECHS1	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
FGF14	99.99 %	601515	Spinocerebellar ataxia 27A, 193003 (3), Autosomal dominant; Spinocerebellar ataxia 27B, late-onset, 620174 (3), Autosomal dominant
FOXG1	99.91 %	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
GABBR2	99.96 %	607340	{Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3); Developmental and epileptic encephalopathy 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant
GABRA1	100 %	137160	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Developmental and epileptic encephalopathy 19, 615744 (3), Autosomal dominant; {Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3)
GCH1	99.94 %	600225	Dystonia, DOPA-responsive, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GLRA1	100 %	138491	Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant
GLRB	99.79 %	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GNAO1	99.86 %	139311	Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
KCNA1	100 %	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNK18	100 %	613655	{Migraine, with or without aura, susceptibility to, 13}, 613656 (3), Autosomal dominant

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KCNMA1	99.89 %	600150	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant
KCNQ2	100 %	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
KCNT1	99.98 %	608167	Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant
NOTCH3	99.99 %	600276	Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant
PDE10A	87.37 %	610652	Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant; Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive
PDE2A	99.95 %	602658	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive
PDHA1	99.04 %	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHX	99.64 %	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PNKD	100 %	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
PRKN	99.99 %	602544	Adenocarcinoma of lung, somatic, 211980 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3)
PRRT2	99.97 %	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant
RHOBTB2	100 %	607352	Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant
SACS	99.97 %	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
SCN1A	99.94 %	182389	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant
SCN2A	99.86 %	182390	Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant
SCN4A	99.98 %	603967	Paramyotonia congenita, 168300 (3), Autosomal dominant; Hyperkalemic periodic paralysis, 170500 (3), Autosomal dominant; Congenital myopathy 22B, severe fetal, 620369 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Congenital myopathy 22A, classic, 620351 (3), Autosomal recessive
SCN8A	99.77 %	600702	?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant
SETX	99.97 %	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SLC16A2	99.97 %	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked

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SLC1A3	99.98 %	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
SLC20A2	99.95 %	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
SLC2A1	99.93 %	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant
SLC6A5	99.97 %	604159	Hyperekplexia 3, 614618 (3), Autosomal recessive, Autosomal dominant
TBC1D24	100 %	613577	Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive
TMEM151A	100 %	620108	Episodic kinesigenic dyskinesia 3, 620245 (3), Autosomal dominant
TPK1	99.96 %	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
TREX1	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
VAMP2	100 %	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* Exome panels: >=20x, HyperCap panels: >=30x