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epiSEEK® Comprehensive Sequence Analysis of Epilepsy and Seizure Disorders (471 genes)

Gene	Disease association	Inheritance
ABAT	GABA-transaminase deficiency	AR
ABCC2	dubin-johnson syndrome	AR
ABCC8	hyperinsulinemic hypoglycemia of infancy	unknown
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR
ACY1	aminoacylase-1 deficiency	AR
ADCK3	Selected inborn errors of metabolism	AR
ADSL	Adenylosuccinase deficiency	AR
AGA	aspartylglycosaminuria	AR
AGTR2	mental retardation, X-linked, 88	XL
AHI1	joubert syndrome-related disorders	AR
AKT3	PPARgamma pathway	Unknown
ALDH4A1	type II hyperpolinemia	AR
ALDH5A1	4-hydroxybutyricaciduria	AR
ALDH7A1	pyridoxine-dependent epilepsy	AR
ALG1	congenital disorder of glycosylation type Ik	AR
ALG11	Congenital disorder of glycosylation	AR
ALG12	congenital disorder of glycosylation type Ig	AR
ALG13	Congenital disorder of glycosylation	XL recessive
ALG2	congenital disorder of glycosylation type Ih	AR
ALG3	congenital disorder of glycosylation type Id	AR
ALG6	congential disorders of glycosylation type Ic	AR
ALG8	congenital disorder of glycosylation type Ih	AR
ALG9	congenital disorder of glycosylation type Il	AR
AMT	glycine encephalopathy	AR
APTX	ataxia-ocular apraxia	AR
ARFGEF2	periventricular hereotopia & microcephaly	AR
ARG1	argininemia	AR
ARHGEF9	startle disease with epilepsy (STHEE)	XL
ARL13B	joubert syndrome 8	AR
ARSA	metachromatic leucodystrophy (MLD)	Unknown
ARSB	mucopolysaccharidosis type VI	AR
ARX	X-linked mental retardation and epilepsy	AR
ASAH1	farber lipogranulomatosis; spinal muscular atrophy with progressive myoclonic epilepsy	AR; AD
ASPA	canavan disease	AR
ASPM	microcephaly primary type 5	AR
ATIC	AICA-ribosiduria	AR
ATN1	denatorubro-pallidoluysian atrophy	AD
ATP1A2	familial basilar or hemiplegi migraines	AD
ATP1A3	alternating hemiplegia of childhood 2; dystonia-12	AD
ATP2A2	darier-white disease	AD
ATP5A1	Mitochondrial complex (ATP synthase) deficiency, nuclear type 4	AR
ATP6AP2	Mental retardation, X-linked, with epilepsy	XL
ATP6V0A2	cutis laxa type II and wrinkly skin syndrome	AR
ATP7A	menkes disease; occiptal horn syndrome; spinal muscular atrophy, distal, X-linked 3	XL
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency	AR
ATR	seckel syndrome	AR
ATRX	X-linked mental retardation (XLMR) syndrome most often accompanied by alpha-thalassemia (ATRX) syndrome	AR
B4GALT1	Congenital disorder of glycosylation	AR
BCKDHA	maple syrup urine disease, type Ia	AR
BCKDHB	maple syrup urine disease, type Ib	AR

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BCKDK	branched-chain ketoacid dehydrogenase kinase deficiency	AR
BCS1L	mitochondrial complex III deficiency and the GRACILE syndrome	AR
BRAF	cardiofaciocutaneous syndrome	unknown
BRAT1	rigidity and multifocal seizure syndrome	AR
BRD2	juvenile myoclonic epilepsy	unknown
BTD	Biotinidase deficency	AR
BUB1B	Mosaic variegated aneuploidy syndrome	AD; AR
C12ORF57	Temtamy syndrome, Joubert syndrome and related disorders	AR
C12ORF65	Mitochondrial complex IV deficiency	AR
CACNA1A	familial hemiplegic migraine and episodic ataxia 2	AD
CACNA1H	childhood absence epilepsy (CAE)	AD
CACNB4	idiopathic generalized epilepsy (IGE) and juvenile myoclonic epilepsy (JME)	AD
CASK	FG syndrome 4, mental retardation and microcephaly with pontine and cerebellar hypoplasia, and a form of X-linked mental retardation	AR
CASR	familial hypocalciuric hypercalcemia, familial, isolated hypoparathyroidism, and neonatal severe primary hyperparathyroidism	AR; AD
CBL	noonan syndrome-like disorder	AD
CC2D2A	meckel syndrome type 6, Joubert syndrome type 9	unknown
CCDC88C	negative regulator of the canonical Wnt signaling pathway	AR
CCL2	coronary artery disease, modifier of; HIV-1, resistance to; myobacterium tuberculosis, susceptibility to; spina bifida, susceptibility to	AR
CDK5RAP2	Microcephaly 3, primary, autosomal recessive	AR
CDKL5	X-linked infantile spasm syndrome (ISSX), X-linked west syndrome, Rett syndrome (RTT)	XL
CDON	holoprosencephaly 11	AD
CENPJ	primary autosomal recessive microcephaly	AR
CEP152	primary microcephaly (MCPH4)	AR
CEP290	joubert syndrome and nephronophthisis	AR
CHD2	epileptic encephalopathy, childhood-onset	AD
CHRNA2	nocturnal frontal lobe epilepsy type 4	AD
CHRNA4	nocturnal frontal lobe epilepsy type 1	AD
CHRNA7	intellectual disabiliy, epilepsy, schizophrenia, autism spectrum disorder	AD
CHRNB2	nocturnal frontal lobe epilepsy	AD
CLCN2	idiopathic generalized epilepsy (IGE), juvenile absence epilepsy (JAE), and juvenile myoclonic epilepsy (JME)	AD
CLCNKA	Bartter syndrome, type 4b	unknown
CLCNKB	bartter syndrome type 3 (BS3)	AR
CLN3	batten disease or neuronal ceroid lipofuscinoses (NCLs)	AR
CLN5	batten disease	AR
CLN6	batten disease	AR
CLN8	progressive epilepsy with mental retardation (EMPR)	AR
CNR1	Cannabinoid receptor 1	Unknown
CNR2	Cannabinoid receptor 2	Unknown
CNTN2	Generalized/Myoclonic/Absence Epilepsies/Febrile Seizures	AR
CNTNAP2	gilles de la tourette syndrome, schizophrenia, epilepsy, autism, ADHD and mental retardation	AR
COG1	Congenital disorder of glycosylation	AR
COG4	Congenital disorder of glycosylation	AR
COG5	Congenital disorder of glycosylation	Likely Recessive
COG6	Congenital disorder of glycosylation	AR
COG7	congenital disorder of glycosylation type IIe	AR

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Gene	Disease association	Inheritance
COG8	congenital disorder of glycosylation, type IIh	unknown
COL18A1	knobloch syndrome	AR
COL4A1	Porencephaly	AD
COQ2	coenzyme Q10 deficiency, COQ2 nephropathy	AR
COQ9	coenzyme Q10 deficiency	AR
COX10	Leigh disease	AR
COX15	Leigh syndrome	AR
CPT1A	CPT deficiency, hepatic type 1A	AR
CPT2	mitochondrial long-chain fatty-acid (LCFA) oxidation disorders	AR
CSTB	progressive myoclonic epilepsy (EPM1)	AR
CTSA	galactosialidosis	unknown
CTSD	Ceroid lipofuscinosis, neurona	AR
CTSF	Kufs Disease	AR
CUL4B	the encoded protein forms a complex that functions as an E3 ubiquitin ligase and catalyzes the polyubiquitination of specific protein substrates	unknown
CYP2C19	cytochrome P450, family 2, subfamily C, polypeptide 19	Complex
CYP2C9	cytochrome P450, family 2, subfamily C, polypeptide 9	AD
CYP3A4	cytochrome P450, family 3, subfamily A, polypeptide 4	Complex
CYP3A5	cytochrome P450, family 3, subfamily A, polypeptide 5	Complex
DAGLA	Endocannabinoid metabolism	Unknown
DBT	maple syrup urine disease, type II	AR
DCLK2	severe epileptic phenotype and lethality, as described in human patients with lissencephaly	unknown
DCX	epilepsy, mental retardation, subcortical band heterotopia ("double cortex" syndrome) in females and lissencephaly ("smooth brain" syndrome) in males	AD
DDC	Aromatic L-amino acid decarboxylase deficiency	AR
DDOST	Congenital disorder of glycosylation	AR
DEPDC5	epilepsy, familial focal, with variable foci	AD
DHCR7	Smith-Lemli-Opitz syndrome	AR
DLD	E3-deficient maple syrup urine disease and lipoamide dehydrogenase deficiency	AR
DLGAP2	suspected in autism spectrum disorder	AR
DNAJC5	Kufs Disease, NCL-4B	AD
DOLK	dolichol kinase deficiency	AR
DPAGT1	congenital disorder of glycosylation type Ij	AR
DPM1	serves as a donor of mannosyl residues on the lumenal side of the endoplasmic reticulum (ER)	AR
DPM3	serves as a donor of mannosyl residues on the lumenal side of the endoplasmic reticulum (ER)	AR
DPYD	dihydropyrimidine dehydrogenase deficiency	AR
DYRK1A	Mental retardation, autosomal dominant 7	AD
EFHC1	juvenile myoclonic epilepsy and juvenile absence epilepsy	unknown
EFHC2	Turner syndrome	XL
EIF2B1	leukoencephalopathy with vanishing white matter	AR
EIF2B2	leukoencephalopathy with vanishing white matter	AR
EIF2B3	leukoencephalopathy with vanishing white matter	AR
EIF2B4	leukoencephalopathy with vanishing white matter	AR
EIF2B5	leukoencephalopathy with vanishing white matter	AR
ELP4	rolandic epilepsy	AR;AD
EMX2	Schizencephaly	AD
EOMES	t-box genes encode transcription factors involved in the regulation of developmental processes	unknown
EPM2A	myoclonic epilepsy of Lafora	AR
ETFA	type II glutaricaciduria	AR
ETFB	type II glutaricaciduria	AR
ETFDH	type II glutaricaciduria	AR

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Gene	Disease association	Inheritance
FAAH	Endocannabinoid metabolism	Unknown
FGD1	faciogenital dysplasia and X-linked mental retardation, syndromatic 16	XL
FGF8	this protein is known to be a factor that supports androgen and anchorage independent growth of mammary tumor cells	AD
FGFR3	craniosynostosis and multiple types of skeletal dysplasia	AD
FH	fumarase deficiency and l progressive encephalopathy	AR
FKRP	congenital muscular dystrophy, mental retardation, and cerebellar cysts	AR
FKTN	fukuyama-type congenital muscular dystrophy (FCMD), walker-warburg syndrome (WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilated cardiomyopathy type 1X (CMD1X)	AR
FLNA	periventricular nodular heterotopias (PVNH1, PVNH4), otopalatodigital syndromes (OPD1, OPD2), frontometaphyseal dysplasia (FMD), melnick-needles syndrome (MNS), and X-linked congenital idiopathic intestinal pseudoobstruction (CIIPX)	AD; AR
FLVCR2	proliferative vaculopathy and hydraencephaly-hydrocephaly syndrome	AR
FOLR1	neurodegeneration due to cerebral folate transport deficiency	AR
FOXG1	rett syndrome	AD
FOXH1	nonsyndromic holoprosencephaly; critical congenital heart disease	AD
FUCA1	fucosidosis (FUCA1D)	AR
GABBR1	schizophrenia; epilepsy; multiple sclerosis	unknown
GABBR2	gamma-aminobutyric acid (GABA) B receptor, 2	Unknown
GABRA1	juvenile myoclonic epilepsy and childhood absence epilepsy type 4	AD
GABRA2	gamma-aminobutyric acid (GABA) A receptor, alpha 2	Unknown
GABRA3	gamma-aminobutyric acid (GABA) A receptor, alpha 3	Unknown
GABRA4	gamma-aminobutyric acid (GABA) A receptor, alpha 4	Unknown
GABRA5	gamma-aminobutyric acid (GABA) A receptor, alpha 5	Unknown
GABRA6	schizophrenia, susceptibility to	AD
GABRB1	gamma-aminobutyric acid (GABA) A receptor, beta 1	Unknown
GABRB2	gamma-aminobutyric acid (GABA) A receptor, beta 2	Unknown
GABRB3	angelman syndrome, Prader-Willi syndrome, and autism	AD
GABRD	generalized epilepsy with febrile seizures, type 5	AD
GABRE	gamma-aminobutyric acid (GABA) A receptor, epsilon	Unknown
GABRG1	alcoholism, susceptibility to	AR;AD
GABRG2	epilepsy and febrile seizures	AD
GABRG3	gamma-aminobutyric acid (GABA) A receptor, gamma 3	Unknown
GABRP	gamma-aminobutyric acid (GABA) A receptor, pi	Unknown
GABRQ	gamma-aminobutyric acid (GABA) A receptor, theta	Unknown
GABRR1	gamma-aminobutyric acid (GABA) A receptor, rho 1	Unknown
GABRR2	gamma-aminobutyric acid (GABA) A receptor, rho 2	Unknown
GABRR3	gamma-aminobutyric acid (GABA) A receptor, rho 3	Unknown
GALC	krabbe disease	AR
GALNS	morquio A syndrome	AR
GAMT	neurologic syndromes and muscular hypotonia	AR
GATM	arginine:glycine amidinotransferase deficiency	AR
GCDH	Glutaricaciduria	AR
GCSH	nonketotic hyperglycinemia (NKH)	AR
GFAP	alexander disease	AD
GJD2	myopia	AR;AD
GLB1	GM1-gangliosidosis and Morquio B syndrome	AR
GLDC	nonketotic hyperglycinemia (NKH)	AR

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Gene	Disease association	Inheritance
GLI2	greig cephalopolysyndactyly syndrome, pallister-hall syndrome, preaxial polydactyly type IV, postaxial polydactyly types A1 and B	AD
GLI3	greig cephalopolysyndactyly syndrome, pallister-hall syndrome, preaxial polydactyly type IV, postaxial polydactyly types A1 and B	AD
GLRA1	startle disease (STHE)	AD; AR
GLRB	startle disease (STHE)	AR
GLUD1	Hyperinsulinism-hyperammonemia syndrome	AD
GLUL	Glutamine deficiency, congenital	AR
GNE	sialuria, autosomal recessive inclusion body myopathy, and Nonaka myopathy	unknown
GNPTAB	mucolipidosis II and mucolipidosis IIIA	AR
GNPTG	mucolipidosis IIIC	AR
GNS	mucopolysaccharidosis type IIID (sanfilippo D syndrome)	AR
GOSR2	Epilepsy, progressive myoclonic 6	AR
GPC3	simpson-golabi-behmel syndrome	XL
GPHN	hyperplexia and molybdenum cofactor deficiency	AR
GPR55	Cannabinoid receptor	Unknown
GPR56	bilateral frontoparietal polymicrogyria	unknown
GPR98	usher syndrome 2 and familial febrile seizures	AD; AR
GRIA3	Mental retardation, X-linked 94	XL
GRIN1	mental retardatiomn, autosomal dominant 8	AD
GRIN2A	N-methyl-D-aspartate (NMDA) receptors are a class of ionotropic glutamate receptors	AD
GRIN2B	N-methyl-D-aspartate (NMDA) receptors are a class of ionotropic glutamate receptors	AD
GUSB	mucopolysaccharidosis type VII	AR
HCN1	epilepsy; sudden unexpected death in epilepsy	likely AR
HCN2	generalized epilepsy; sudden unexpected death in epilepsy	AR
HCN3	sudden unexpected death in epilepsy	likely AR
HCN4	brugada syndrome 8; sick sinus syndrome 2; sudden unexpected death in epilepsy	AD
HERC2	Mental retardation, autosomal recessive 38	AR
HEXA	tay-sachs disease (GM2-gangliosidosis type I)	AR
HEXB	sandhoff disease (GM2-gangliosidosis type II)	unknown
HGSNAT	sanfilippo syndrome C	AR
HNRNPU	Epileptic encephalopathy	AD
HPD	tyrosinemia type 3 (TYRO3) and hawkinsinuria (HAWK)	AD
HRAS	costello syndrome	AD
HSD17B10	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (MHBD)	unknown
HSD17B4	d-bifunctional protein deficiency; perrault syndrome 1	AR
HYAL1	mucopolysaccharidosis type IX, or hyaluronidase deficiency	AR
IDH2	d-2-hydroxygluaric aciduria 2	AD
IDS	mucopolysaccharidosis type II, also known as hunter syndrome	AR
IDUA	mucopolysaccharidosis type I (MPS I)	AR
INPP5E	joubert syndrome	AR
IQSEC2	Epilepsy in X-linked intellectual disability	XL
JRK	childhood absence epilepsy	AR
KANSL1	Koolen-de Vries syndrome	AD
KAT6B	Malformation disorders including neuronal migration disorders, severe microcephaly, pontocerebellar hypoplasia, Joubert syndrome and related disorders, holoprosencephaly, and disorders of the RAS/MAPK pathway	AD
KCNA1	myokymia with periodic ataxia (AEMK)	AD
KCNAB1	schizophrenia	unknown
KCNJ1	antenatal bartter syndrome	AR
KCNJ10	seizure susceptibility of common idiopathic generalized epilepsy syndromes	AR

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Gene	Disease association	Inheritance
KCNJ11	ATP-binding mutation	AD
KCNMA1	generalized epilepsy and paroxysmal dyskinesia	AD
KCNQ2	benign familial neonatal convulsions type 1 (BFNC), also known as epilepsy, benign neonatal type 1 (EBN1)	AD
KCNQ3	benign familial neonatal convulsions type 2 (BFNC2), also known as epilepsy, benign neonatal type 2 (EBN2)	AD
KCNT1	epilepsy, nocturnal frontal lobe, 5; epileptic encephalopathy, early infantile, 14	AD
KCNV2	retinal cone dystrophy 3B	AR
KCTD7	progressive myoclonic epilepsy-3	AR
KDM5C	X-linked mental retardation	XL
KIAA1279	goldberg-shprintzen megacolon syndrome	AR
KMT2D	kabuki syndrome	AD
KRAS	noonan syndrome, cardiofaciocutaneous, encodes a protein that is a member of the small GTPase superfamily	AD
L2HGDH	L-2-hydroxyglutaric aciduria	AR
LAMA2	congenital merosin-deficient muscular dystrophy	AR
LARGE	MDC1D	unknown
LBR	HEM/greenberg skeletal dysplasia	AR
LGI1	lateral temporal epilepsy	AD
LIAS	pyruvate dehydrogenase lipoic acid synthetase deficiency	AR
LIG4	LIG4 syndrome	AR
LRPPRC	french-canadian Leigh disease	AR
MAGI2	Infantile spasms is associated with deletion of the MAGI2	AR;AD
MAGT1	immunodeficiency, X-linked, with magnesium defect, epstein-barr virus infection and neoplasia; mental retardation, x-linked 95	XL
MAP2K1	Cardiofaciocutaneous syndrome 3	AD
MAP2K2	cardiofaciocutaneous syndrome (CFC syndrome)	AD
MAPK10	this kinase plays regulatory roles in the signaling pathways during neuronal apoptosis	AD
MBD5	mental retardation autosomal dominant type 1	AD
MCOLN1	mucolipidosis type IV	AR
MCPH1	primary autosomal recessive microcephaly 1 and premature chromosome condensation syndrome	AR
ME2	Epilepsy, idiopathic generalized	unknown
MECP2	rett syndrome	XL
MED12	lujan-fryns syndrome; ohdo syndrome, X-linked; opitz-kaveggia syndrome	XL
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR
MEF2C	chromosome 5q14.3 deletion syndrome; mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	AD
MFSD8	variant form of late infantile-onset neuronal ceroid lipofuscinoses (vLINCL)	AR
MGAT2	carbohydrate-deficient glycoprotein syndrome, type II Megalencephalic leukoencephalopathy with subcortical c	AR
MGLL	Endocannabinoid metabolism	Unknown
MGME1	Mitochondrial DNA depletion syndrome	AR
MLC1	megalencephalic leukoencephalopathy with subcortical cysts	AR
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR
MOCS1	molybdenum cofactor deficiency, type A	AR
MOCS2	molybdenum	AR
MOGS	type IIb congenital disorder of glycosylation (CDGIIb)	AR
MPDU1	congenital disorder of glycosylation type If	AR
MPI	carbohydrate-deficient glycoprotein syndrome, type Ib	AR

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Gene	Disease association	Inheritance
MTHFR	homocystinuria due to MTHFR deficiency; neural tube defects, susceptibility to; schizophrenia, susceptibility to; thromboembolism, susceptibility to; vascular disease, susceptibility to	AR
MTOR	PPARgamma pathway	unknown
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	AR
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	AR
NAGLU	mucopolysaccharidosis type IIIB (MPS-IIIB), also known as Sanfilippo syndrome B	AR
NDE1	lissencephaly 4 (with microcephaly)	AR
NDUFA1	mitochondrial complex 1 deficiency	AR
NDUFA2	Leigh disease	AR
NDUFAF6	Selected inborn errors of metabolism	AR
NDUFS1	Leigh disease	AR
NDUFS3	Leigh disease	AR
NDUFS4	Leigh disease	AR
NDUFS7	Leigh disease	AR
NDUFS8	Leigh disease	AR
NDUFV1	Leigh disease	AR
NEDD4L	Epileptic encephalopathy	AD
NEU1	sialidosis	unknown
NF1	neurofibromatosis type 1, juvenile myelomonocytic leukemia and watson syndrome	AD
NGLY1	congenital disorder of glycosylation, type IV	AR
NHEJ1	severe combined immunodeficiency disorders	AR
NHLRC1	lafora disease or progressive myoclonic epilepsy type 2 (EPM2)	AR
NIPBL	cornelia de lange syndrome	AD
NODAL	heterotaxy, visceral, 5	AD
NOTCH3	cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)	AD
NPC1	niemann-pick type C disease	AR
NPC2	niemann-pick disease, type C2 and frontal lobe atrophy	AR
NPHP1	familial juvenile nephronophthisis type 1, senior-loken syndrome type 1, joubert syndrome type 4	unknown
NRAS	Noonan syndrome	AD
NRXN1	neurexins function in the vertebrate nervous system as cell adhesion molecules and receptors	AR; AD
OFD1	oral-facial-digital syndrome type I and simpson-golabi-nbehmel syndrome type 2	AR
OPA1	optic atrophy 1; optic atrophy plus syndrome; glaucoma, normal tension, susceptibility to	AD
OPHN1	X-linked mental retardation	XL
PAFAH1B1	miller-dieker lissencephaly syndrome	AD
PAK3	non-syndromic mental retardation X-linked type 30 (MRX30)	XL
PANK2	HARP syndrome and pantothenate kinase-associated neurodegeneration (PKAN)	AR
PAX6	ocular disorders such as aniridia and peter's anomaly	unknown
PC	pyruvate carboxylase deficiency	AR
PCDH19	epilepsy female-restricted with mental retardation (EFMR)	XL
PCNT	seckel syndrome-4 and microcephalic osteodysplastic primordial dwarfism type II	AR
PDHA1	pyruvate dehydrogenase E1-alpha deficiency and X-linked Leigh disease	XL
PDHX	pyruvate dehydrogenase deficiency	AR
PDSS1	coenzyme Q10 deficiency	AR
PDSS2	coenzyme Q10 deficiency	AR
PEX1	complementation group 1 peroxisomal disorders such as neonatal adrenoleukodystrophy, infantile Refsum disease, and zellweger syndrome	AR
PEX10	Zellweger syndrome, Peroxisome biogenesis disorder	likely AR
PEX12	zellweger syndrome (ZWS)	AR
PEX13	Zellweger syndrome, Peroxisome biogenesis disorder	likely AR

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PEX14	zellweger syndrome	AR
PEX16	Zellweger syndrome, Peroxisome biogenesis disorder	likely AR
PEX19	Zellweger syndrome, Peroxisome biogenesis disorder	likely AR
PEX2	zellweger syndrome and infantile refsum disease	AR
PEX26	zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP)	AR
PEX3	zellweger syndrome (ZWS)	AR
PEX5	neonatal adrenoleukodystrophy (NALD), a cause of zellweger syndrome (ZWS) as well as may be a cause of infantile refsum disease (IRD)	AR
PEX6	Zellweger syndrome, Peroxisome biogenesis disorder	AR
PEX7	PBD complementation group 11 (PBD-CG11) disorders, rhizomelic chondrodysplasia punctata type 1 (RCDP1), and refsum disease (RD)	AR
PGK1	Phosphoglycerate kinase 1 deficiency	XL
PGM1	Zellweger syndrome, Peroxisome biogenesis disorder	AR
PHF6	borjeson-forssman-lehmann syndrome (BFLS)	unknown
PHGDH	phosphoglycerate dehydrogenase deficiency	AR
PIGV	hyperphosphatasia mental retardation syndrome	AR
PIK3CA	megalencephaly-capillary malformation syndrome	Somatic mutation
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	Somatic mutation
PLA2G6	Neurodegeneration with brain iron accumulation, Infantile neuroaxonal dystrophy	AR
PLCB1	epileptic encephalopathy, early infantile, 12	AR
PLP1	X-linked pelizaeus-merzbacher disease and spastic paraplegia type 2	AR
PMM2	carbohydrate-deficient glycoprotein syndrome type I	AR
PNKP	microcephaly, seizures, and developmental delay	AR
PNPO	pyridoxamine 5'-phosphate oxidase (PNPO) deficiency	AR
POLG	progressive external ophthalmoplegia with mitochondrial DNA deletions 1 (PEOA1), sensory ataxic neuropathy dysarthria and ophthalmoparesis (SANDO), alpers-huttenlocher syndrome (AHS), and mitochondrial neurogastrointestinal encephalopathy syndrome (MNGIE)	AD; AR
POMGNT1	muscle-eye-brain (MEB) disease	AR
POMT1	walker-warburg syndrome (WWS) and limb-girdle muscular dystrophy type 2K (LGMD2K)	AR
POMT2	walker-warburg syndrome (WWS)	AR
PPT1	infantile neuronal ceroid lipofuscinosis 1 (CLN1, or INCL) and neuronal ceroid lipofuscinosis 4 (CLN4)	AR
PQBP1	renpenning syndrome 1 and other syndromes with X-linked mental retardation	XL
PRICKLE1	progressive myoclonus epilepsy	AR; AD
PRICKLE2	progressive myoclonic epilepsy type 5	AD
PRODH	hyperprolinemia type 1 and susceptibility to schizophrenia 4 (SCZD4)	AR
PRRT2	convulsions, familial infantile, with paroxysmal choreothetosis; episodic kinesigenic dyskinesia 1; seizures, benign familial infantile, 2	AD
PSAP	gaucher disease, tay-sachs disease, and metachromatic leukodystrophy	AR
PSAT1	Phosphoserine aminotransferase deficiency	AR
PTCH1	basal cell nevus syndrome, esophageal squamous cell carcinoma, trichoepitheliomas, transitional cell carcinomas of the bladder, and holoprosencephaly	likely AD
PTPN11	noonan syndrome and acute myeloid leukemia	AD
QDPR	atypical phenylketonuria	AR
RAB39B	rab proteins are small GTPases that are involved in vesicular trafficking	unknown
RAB3GAP1	warburg micro syndrome	unknown
RAF1	noonan syndrome 5 and LEOPARD syndrome 2	unknown
RAI1	Smith-Magenis syndrome	AD
RARS2	pontocerebellar hypoplasia type 6 (PCH6)	AR

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RBFOX1	spinocerebellar ataxia type 2 (SCA2)	unknown
RELN	autosomal recessive lissencephaly with cerebellar hypoplasia	AR
RFT1	glycosylation disorder type In	likely AR
RNASEH2A	aicardi-goutieres syndrome (AGS)	unknown
RNASEH2B	aicardi-goutieres syndrome type 2 (AGS2)	AR
RNASEH2C	aicardi-goutieres syndrome-3	AR
RPGRIP1L	joubert syndrome type 7 (JBTS7) and meckel syndrome type 5 (MKS5)	AR
RTTN	polymicrogyria with seizures	AR
SAMHD1	aicardi-goutieres syndrome	AR
SCARB2	autosomal recessive progressive myoclonic epilepsy-4 (EPM4)	AR
SCN10A	small fiber neuropathy	unknown
SCN11A	Neuropathy, hereditary sensory and autonomic, type VII	unknown
SCN1A	dravet syndrome; epilepsy, generalized, with febrile seizures plus, type 2; febrile seizures, familial, 3A; migraine, familial hemiplegic, 3	AD
SCN1B	generalized epilepsy with febrile seizure, brugada syndrome 5, and defects in cardiac conduction	AD
SCN2A	several seizure disorders	AD
SCN2B	Atrial fibrillation, familial, 14	AD
SCN3A	No known disease association	unknown
SCN3B	brugada syndrome 7	AD
SCN4A	hyperkalemic periodic paralysis, type 2; hypokalemic periodic paralysis, type 2; myasthenic syndrome, acetazolamide-responsive; myotonia congenita, atypical, acetazolamide-responsive; paramyotonia congenita	AD
SCN4B	long QT syndrome type 10 (LQT10).	AD
SCN5A	long QT syndrome type 3 (LQT3)	AD
SCN7A	neonatal seizures	AD
SCN8A	mental retardation, pancerebellar atrophy and ataxia	AD
SCN9A	primary erythermalgia, channelopathy-associated insensitivity to pain, and paroxysmal extreme pain disorder	AD
SCO2	fatal infantile encephalocardiomyopathy	AR
SDHA	Leigh disease	AR
SERPINI1	familial encephalopathy with neuroserpin inclusion bodies (FENIB)	AD
SETBP1	schinzel-giedion midface retraction syndrome	AD
SGCE	dystonia-11, myoclonic	AD
SGSH	sanfilippo syndrome A	AR
SHH	holoprosencephaly (HPE)	AD
SHOC2	noonan-like syndrome with loose anagen hair	unknown
SIX3	holoprosencephaly type 2	AD
SLC16A2	allan-herndon-dudley syndrome	unknown
SLC17A5	sialic acid storage diseases	AR
SLC19A3	Thiamine metabolism dysfunction syndrome 2	AR
SLC1A3	episodic ataxia	unknown
SLC25A15	hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	AR
SLC25A19	microcephaly, amish type	AR
SLC25A22	early infantile epileptic encephalopathy	AR
SLC2A1	paroxysmal exertion-induced dyskinesia	AD; AR
SLC35A1	congenital disorder of glycosylation type 2F (CDG2F)	AR
SLC35A2	Congenital disorder of glycosylation	XL dominant
SLC35C1	congenital disorder of glycosylation type IIc	AR
SLC46A1	autosomal recessive hereditary folate malabsorption (HFM) disease	AR
SLC4A10	None Known	unknown

epiSEEK® Comprehensive Sequence Analysis of Epilepsy and Seizure Disorders (471 genes)

Gene	Disease association	Inheritance
SLC6A4	Serotonin transporter deficiency, Epileptic encephalopathy	AD
SLC6A5	Hyperekplexia	AR; AD
SLC6A8	cerebral creatine deficiency syndrome 1	XL
SLC9A6	X-linked syndromic mental retardation, Christianson type	XL
SMC1A	Cornelia de Lange syndrome 2	XL dominant
SMC3	Cornelia de Lange syndrome 3	unknown
SMPD1	niemann-pick disease type A (NPA) and niemann-pick disease type B (NPB)	AR
SMS	X-linked snyder-robinson mental retardation syndrome	XL
SNAP25	Neuropsychiatric and Neurological Disorders	AD
SNAP29	Cerebral dysgenesis, neuropathy	AR
SNIP1	psychomotor retardation, epilepsy, and craniofacial dysmorphism	AR
SOS1	gingival fibromatosis 1 and noonan syndrome type 4	AD
SPRED1	neurofibromatosis type 1-like syndrome (NFLS)	AD
SPTAN1	early infantile epileptic encephalopathy-5	AD
SRD5A3	Congenital disorder of glycosylation	AR
SRPX2	bilateral perisylvian polymicrogyria, rolandic epilepsy, speech dyspraxia and mental retardation	unknown
ST3GAL3	epileptic encephalopathy, early infantile, 15; mental retardation, autosomal recessive 12	AR
ST3GAL5	amish infantile epilepsy syndrome	AR
STIL	Microcephaly 7, primary, autosomal recessive	AR
STRADA	polyhydramnios, megalencephaly, and symptomatic epilepsy	AR
STXBP1	infantile epileptic encephalopathy-4	AD
SUCLA2	Mitochondrial DNA depletion syndrome	AR
SUMF1	multiple sulfatase deficiency, a lysosomal storage disorder	AR
SUOX	sulfite oxidase deficiency	AR
SURF1	Leigh disease	AR
SYN1	X-linked disorders with primary neuronal degeneration such as rett syndrome	XL
SYNGAP1	mental retardation autosomal dominant type 5	AD
SYP	X-linked mental retardation (XLMR)	XL
TACO1	Leigh disease	AR
TBC1D24	familial infantile myoclonic epilepsy	AR
TBX1	Velocardiofacial syndrome, DiGeorge syndrome	AD
TCF4	pitt-hopkins syndrome	AD; AR
TGIF1	holoprosencephaly type 4	AD
TMEM165	Congenital disorder of glycosylation, type IIk	AR
TMEM216	meckel-gruber syndrome type 2, and joubert syndrome 2	AR
TMEM67	meckel syndrome type 3 (MKS3) and joubert syndrome type 6 (JBTS6)	AR
TMEM70	neonatal mitochondrial encephalocardiomyopathy	AR
TPP1	late-infantile neuronal ceroid lipofuscinosis	AR
TREX1	aicardi-goutieres syndrome, chilblain lupus, cree encephalitis, and other diseases of the immune system	AD; AR
TRPM6	hypomagnesemia 1, intestinal	AR
TSC1	tuberous sclerosis	AD
TSC2	tuberous sclerosis complex	AD
TSEN2	pontocerebellar hypoplasia type 2	AR
TSEN34	pontocerebellar hypoplasia type 2	AR
TSEN54	pontocerebellar hypoplasia type 2	AR
TUBA1A	lissencephaly type 3 (LIS3)	AD
TUBA8	polymicrogyria and optic nerve hypoplasia	AR
TUBB2B	asymmetric polymicrogyria	AD
TUSC3	Mental retardation, autosomal recessive 7	AR
UBE3A	angelman syndrome	imprinted

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Gene	Disease association	Inheritance
VANGL1	caudal regression syndrome; neural tube defects	AD
VDAC1	the encoded protein facilitates the exchange of metabolites and ions across the outer mitochondrial membrane and may regulate mitochondrial functions	likely AR
VPS13A	cohen syndrome	AR
VPS13B	cohen syndrome	AR
VRK1	pontocerebellar hypoplasia	AR
WDR62	microencephaly, cortical malformations, and mental retardation	AR
ZEB2	hirschsprung disease/mowat-wilson syndrome	AD
ZIC2	holoprosencephaly type 5	AD