iGen
Tegevusluba L02423

Epilepsia NGS paneel

Üldine info

Analüüsi kirjeldus: Epilepsiaga seotud geenide uurimine. Uuritakse 203 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ABAT, ACTL6B, ADAM22, ADAR, ADPRHL2, ADSL, ALDH7A1, ALG13, AMT, AP2M1, AP3B2, APOPT1, ARHGEF9, ARX, ASNS, ATP6V1A, BRAT1, CACNA1A, CACNA1B, CACNA1E, CASK, CDKL5, CHD2, CLCN4, CLTC, CNKSR2, CNPY3, CNTNAP2, COX6B1, CPT2, CYFIP2, D2HGDH, DCX, DENND5A, DNM1, DNM1L, DOCK7, ECHS1, EEF1A2, ETHE1, FAR1, FARS2, FGF12, FLNA, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GPHN, GRIN1, GRIN2A, GRIN2B, GTPBP3, HCN1, HECW2, HEPACAM, HIBCH, HNRNPU, HTT, KCNA2, KCNB1, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KIF1A, LRPPRC, LYRM7, MBD5, MDH2, MECP2, MED17, MEF2C, MOCS1, MRPL44, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTFMT, MTHFR, NACC1, NDUFAF6, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEUROD2, NRXN1, NUBPL, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLAA, PLCB1, PNKP, PNPO, POLG, PPP3CA, PROSC, PTPN23, PURA, RMND1, RNASEH2A, RNASEH2B, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO1, SDHAF1, SERAC1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A1, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SNAP25, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TBCK, TCF4, TRAK1, TREX1, TRIM8, TSC1, TSC2, TTC19, UBA5, UBE3A, UNC80, VARS, WARS2, WDR45, WWOX, ZEB2*

Haigekassa kood: 66618x4

Meetod: 203 geenide kõiki kodeerivaid eksoneid ja ekson-intron piirialasid uuritakse järgmise põlvkonna sekveneerimismeetodiga (NGS, Illumina).

Analüüsi vastus: Analüüsil määratakse, kas uuritav proov on:

a. ***Wild type* ehk metsik-tüüpi (mutatsioone ei esine)**
b. **Mutant (esineb mutatsioon)**

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Kasutatav metoodika ei võimalda uurida harva esinevaid suuri deletsioone ja duplikatsioone ning mutatsioone, mis paiknevad praimeri regioonis või väljaspool fragmenti, mida analüüsitakse.

Logistika

Uuritav proov: Täisveri (EDTA katsutis, lilla korgiga), 2-4 ml.

Kriteeriumid proovile: Vereproovi mitte külmutada, soovitatavalt hoida +4°C juures.

Tellimine: Proovi valmisolekul helistada telefonile **6000 199** ja labor korraldab proovi transpordi. Palun veenduda, et uuringusse saadetav proov on selgelt märgistatud ja lisatud on **saatekiri**.

Teostamise aeg: kuni 4 nädalat

Näidustused: Epilepsia (epileptilised entsefalopaatiad) on närvisüsteemi krooniline haigus, mille tunnuseks on korduvad ja mitteprovotseeritud epileptilised hood.

Epileptiliste entsefalopaatiate etioloogia on erinev ja hõlmab arenguhäireid, metaboolseid haigusi või ka geneetilisi põhjusi Sõltuvalt spetsiifilisest sündroomist ja põhjuslikust geenist võib epilepsia entsefalopaatia olla pärilik autosomaalretsessiivsel, autosomaaldominantsel või X-liiteliselt. Sagedased on samas de novo mutatsioonid.

Geneetiline testimine on näidustatud pärilike epileptiliste entsefalopaatiate diferentsiaaldiagnostikas, sünnieelses diagnostikas ning geneetiliseks konsultatsiooniks geneetilise etioloogiaga peredes.

Geenide nimekiri (203):

Geen	Fenotüüp
ABAT	GABA-transaminase deficiency
ACTL6B	Epilepitic encephalopathy
ADAM22	Early infantile epileptic encephalopathy
ADAR	Dyschromatosis symmetrica hereditaria, Aicardi-Goutières syndrome
ADPRHL2	Neurodegeneration, childhood-onset, with brain atrophy

ADSL	Adenylosuccinase deficiency
ALDH7A1	Epilepsy, pyridoxine-dependent
ALG13	Congenital disorder of glycosylation
AMT	Glycine encephalopathy
AP2M1	Epilepitic encephalopathy
AP3B2	Epileptic encephalopathy, early infantile, 48
APOPT1	Mitochondrial complex IV deficiency
ARHGEF9	Epileptic encephalopathy, early infantile
ARX	Lissencephaly, Epileptic encephalopathy, Corpus callosum, agenesis of, with abnormal genitalia, Partington syndrome, Proud syndrome, Hydranencephaly with abnormal genitalia, Mental retardation
ASNS	Asparagine synthetase deficiency
ATP6V1A	Cutis laxa, autosomal recessive, type IID, Epileptic encephalopathy
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal
CACNA1A	Migraine, familial hemiplegic, Episodic ataxia, Spinocerebellar ataxia 6, Epileptic encephalopathy, early infantile, 42
CACNA1B	Dystonia 23, Early infantile epileptic encephalopathy
CACNA1E	Epileptic encephalopathy
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, FG syndrome, Mental retardation
CDKL5	Epileptic encephalopathy, early infantile, Rett syndrome, atypical, Angelman-like syndrome
CHD2	Epileptic encephalopathy, childhood-onset
CLCN4	Mental retardation, X-linked 49
CLTC	
CNKSR2	Epileptic encephalopathy, X-linked mental retardation, Epilepsy and X-linked mental retardation
CNPY3	Epileptic encephalopathy
CNTNAP2	Pitt-Hopkins like syndrome, Cortical dysplasia-focal epilepsy syndrome
COX6B1	Mitochondrial complex IV deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CYFIP2	Early infantile epileptic encephalopathy, Epilepsy
D2HGDH	D-2-hydroxyglutaric aciduria 1
DCX	Lissencephaly, Subcortical laminal heterotopia
DENND5A	Epileptic encephalopathy, early infantile, 49
DNM1	Epileptic encephalopathy, early infantile
DNM1L	Encephalopathy due to defective mitochondrial and peroxisomal fission 1
DOCK7	Epilepitic encephalopathy

ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
EEF1A2	Epileptic encephalopathy, early infantile, Mental retardation
ETHE1	Ethylmalonic encephalopathy
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder
FARS2	Combined oxidative phosphorylation deficiency 14, Spastic paraplegia 77, autosomal recessive
FGF12	Epileptic encephalopathy, early infantile, 47
FLNA	Frontometaphyseal dysplasia, Osteodysplasty Melnick-Needles, Otopalatodigital syndrome type 1, Otopalatodigital syndrome type 2, Terminal osseous dysplasia with pigmentary defects
FOXG1	Rett syndrome, congenital variant
FRRS1L	Epileptic encephalopathy, early infantile, 37
GABBR2	Epileptic encephalopathy
GABRA1	Epileptic encephalopathy, early infantile, Epilepsy, childhood absence, Epilepsy, juvenile myoclonic
GABRB2	Epileptic encephalopathy
GABRB3	Epilepsy, childhood absence
GABRG2	Generalized epilepsy with febrile seizures plus, Familial febrile seizures, Dravet syndrome, Epilepsy, childhood absence
GAMT	Guanidinoacetate methyltransferase deficiency
GLDC	Glycine encephalopathy
GNAO1	Epileptic encephalopathy, early infantile, Epileptic encephalopathy, early infantile, 17
GPHN	Hyperekplexia, Molybdenum cofactor deficiency
GRIN1	Neurodevelopmental disorder, Mental retardation, autosomal dominant 8
GRIN2A	Epilepsy, focal, with speech disorder
GRIN2B	Epileptic encephalopathy, early infantile, Mental retardation
GTPBP3	Combined oxidative phosphorylation deficiency 23
HCN1	Epileptic encephalopathy, early infantile
HECW2	Neurodevelopmental disorder with hypotonia, seizures, and absent language
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts, remitting
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HNRNPU	Intellectual disability and seizures
HTT	Huntington disease, Lopes-Maciel-Rodan syndrome (LOMARS)
KCNA2	Epileptic encephalopathy, early infantile
KCNB1	Early infantile epileptic encephalopathy
KCNMA1	Paroxysmal nonkinesigenic dyskinesia 3 with or without generalized epilepsy (PNKD3), Cerebellar atrophy, developmental delay, and seizures (CADEDS)

KCNQ2	Epileptic encephalopathy, early infantile, Benign familial neonatal seizures, Myokymia
KCNQ3	Seizures, benign neonatal
KCNQ5	Mental retardation, autosomal dominant 46
KCNT1	Epilepsy, nocturnal frontal lobe
KCNT2	Epileptic encephalopathy
KCTD3	Epileptic encephalopathy
KIF1A	Spastic paraplegia, Neuropathy, hereditary sensory, Mental retardation
LRPPRC	Leigh syndrome, French-Canadian type
LYRM7	Mitochondrial complex III deficiency, nuclear type 8
MBD5	Mental retardation
MDH2	Epileptic encephalopathy, early infantile, 51
MECP2	Angelman-like syndrome, Autism, Rett syndrome, Encephalopathy, Mental retardation
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
MEF2C	Mental retardation
MOCS1	Molybdenum cofactor deficiency
MRPL44	Combined oxidative phosphorylation deficiency 16
MT-ATP6	Neuropathy, ataxia, and retinitis pigmentosa, Leber hereditary optic neuropathy, Ataxia and polyneuropathy, adult-onset, Cardiomyopathy, infantile hypertrophic, Leigh syndrome, Striatonigral degeneration, infantile, mitochondrial
MT-ATP8	Cardiomyopathy, apical hypertrophic, and neuropathy, Cardiomyopathy, infantile hypertrophic
MT-CO1	Myoglobinuria, recurrent, Leber hereditary optic neuropathy, Sideroblastic anemia, Cytochrome C oxidase deficiency
MT-CO2	Cytochrome c oxidase deficiency
MT-CO3	Cytochrome c oxidase deficiency, Leber hereditary optic neuropathy
MT-CYB	
MT-ND1	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, Leber hereditary optic neuropathy, Leber optic atrophy and dystonia
MT-ND2	Leber hereditary optic neuropathy, Mitochondrial complex I deficiency
MT-ND3	Leber optic atrophy and dystonia, Mitochondrial complex I deficiency
MT-ND4	Leber hereditary optic neuropathy, Leber optic atrophy and dystonia, Mitochondrial complex I deficiency
MT-ND4L	Leber hereditary optic neuropathy

MT-ND5	Myoclonic epilepsy with ragged red fibers, Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, Leber hereditary optic neuropathy, Mitochondrial complex I deficiency
MT-ND6	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, Oncocytoma, Leber hereditary optic neuropathy, Leber optic atrophy and dystonia, Mitochondrial complex I deficiency
MT-RNR1	Deafness, mitochondrial
MT-RNR2	Chloramphenicol toxicity/resistance
MT-TA	
MT-TC	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes
MT-TD	
MT-TE	Diabetes-deafness syndrome, Mitochondrial myopathy, infantile, transient, Mitochondrial myopathy with diabetes
MT-TF	Myoclonic epilepsy with ragged red fibers, Nephropathy, tubulointerstitial, Encephalopathy, mitochondrial, Epilepsy, mitochondrial, Myopathy, mitochondrial, Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes
MT-TG	
MT-TH	
MT-TI	
MT-TK	
MT-TL1	Cytochrome c oxidase deficiency, Myoclonic epilepsy with ragged red fibers, Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, Diabetes-deafness syndrome, Cyclic vomiting syndrome, SIDS, susceptibility to
MT-TL2	Mitochondrial multisystemic disorder, Progressive external ophthalmoplegia
MT-TM	Leigh syndrome, Mitochondrial multisystemic disorder
MT-TN	Progressive external ophthalmoplegia, Mitochondrial multisystemic disorder
MT-TP	
MT-TQ	Mitochondrial multisystemic disorder
MT-TR	Encephalopathy, mitochondrial
MT-TS1	Myoclonic epilepsy with ragged red fibers, Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes
MT-TS2	Mitochondrial multisystemic disorder
MT-TT	
MT-TV	Hypertrophic cardiomyopathy (HCM), Leigh syndrome, Mitochondrial multisystemic disorder

MT-TW	Leigh syndrome, Myopathy, mitochondrial
MT-TY	Mitochondrial multisystemic disorder
MTFMT	Combined oxidative phosphorylation deficiency 15
MTHFR	Homocystinuria due to MTHFR deficiency
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination (NECFM)
NDUFAF6	Mitochondrial complex I deficiency, Leigh syndrome
NDUFS2	Mitochondrial complex I deficiency
NDUFS4	Mitochondrial complex I deficiency, Leigh syndrome
NDUFS6	Mitochondrial complex I deficiency
NDUFS7	Mitochondrial complex I deficiency, Leigh syndrome
NDUFS8	Mitochondrial complex I deficiency, Leigh syndrome
NDUFV1	Mitochondrial complex I deficiency
NECAP1	Epileptic encephalopathy, early infantile
NEUROD2	Epileptic encephalopathy
NRXN1	Pitt-Hopkins like syndrome, Developmental delay with or without dysmorphic facies and autism
NUBPL	Mitochondrial complex I deficiency
PARS2	Alpers syndrome
PCDH19	Epileptic encephalopathy, early infantile
PHACTR1	Epileptic encephalopathy
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome
PIGB	Epileptic encephalopathy
PIGP	Epileptic encephalopathy, early infantile, 55
PIGQ	Epileptic encephalopathy
PIGS	Epileptic encephalopathy
PLAA	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies (NDMSBA)
PLCB1	Epileptic encephalopathy, early infantile
PNKP	Epileptic encephalopathy, early infantile, Ataxia-oculomotor
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POLG	POLG-related ataxia neuropathy spectrum disorders, Sensory ataxia, dysarthria, and ophthalmoparesis, Alpers syndrome, Progressive external ophthalmoplegia with mitochondrial DNA deletions, Mitochondrial DNA depletion syndrome
PPP3CA	Epilepitic encephalopathy
PROSC	Epilepsy
PTPN23	Epileptic encephalopathy
PURA	Mental retardation
RMND1	Combined oxidative phosphorylation deficiency
RNASEH2A	Aicardi-Goutières syndrome

RNASEH2B	Aicardi-Goutières syndrome
ROGDI	Kohlschutter-Tonz syndrome
SAMHD1	Aicardi-Goutières syndrome, Chilblain lupus 2
SCN1A	Migraine, familial hemiplegic, Epileptic encephalopathy, early infantile, Generalized epilepsy with febrile seizures plus, Early infantile epileptic encephalopathy 6, Generalized epilepsy with febrile seizures plus, type 2 , Febrile seizures, familial 3A
SCN1B	Atrial fibrillation, Brugada syndrome, Generalized epilepsy with febrile seizures plus, Epilepsy, generalized, with febrile seizures plus, type 1, Epileptic encephalopathy, early infantile, 52
SCN2A	Epileptic encephalopathy, early infantile, Seizures, benign familial infantile
SCN3A	Epilepsy, Epileptic encephalopathy
SCN8A	Cognitive impairment, Epileptic encephalopathy, early infantile
SCO1	Mitochondrial complex IV deficiency
SDHAF1	Mitochondrial complex II deficiency
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome
SIK1	Epileptic encephalopathy, early infantile
SLC12A5	Epileptic encephalopathy, early infantile
SLC13A5	Epileptic encephalopathy, early infantile
SLC19A3	Thiamine metabolism dysfunction syndrome
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria
SLC25A22	Epileptic encephalopathy, early infantile
SLC2A1	Stomatin-deficient cryohydrocytosis with neurologic defects, Epilepsy, idiopathic generalized, GLUT1 deficiency syndrome
SLC35A2	Congenital disorder of glycosylation
SLC6A8*	Creatine deficiency syndrome
SLC9A6	Mental retardation, syndromic, Christianson
SNAP25	Myasthenic syndrome, congenital
SPTAN1	Epileptic encephalopathy, early infantile
ST3GAL3	Epileptic encephalopathy, early infantile, Mental retardation
ST3GAL5	Ganglioside GM3 synthase deficiency
STXBP1	Epileptic encephalopathy, early infantile
SYN1	Epilepsy, with variable learning disabilities and behavior disorders
SYNGAP1	Mental retardation
SYNJ1	Epileptic encephalopathy, early infantile, 53, Parkinson disease 20, early-onset
SZT2	Epileptic encephalopathy, early infantile

TBC1D24	Deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures (DOORS) syndrome, Deafness, autosomal dominant, 65, Myoclonic epilepsy, infantile, familial, Epileptic encephalopathy, early infantile, 16, Deafness, autosomal recessive 86
TBCD	Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum (PEBAT)
TBCE	Progressive encephalopathy with amyotrophy and optic atrophy (PEAMO)
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3
TCF4	Corneal dystrophy, Fuchs endothelial, Pitt-Hopkins syndrome
TRAK1	Epileptic encephalopathy
TREX1	Vasculopathy, retinal, with cerebral leukodystrophy, Chilblain lupus, Aicardi-Goutières syndrome
TRIM8	Epileptic encephalopathy
TSC1	Lymphangioleiomyomatosis, Tuberous sclerosis
TSC2	Lymphangioleiomyomatosis, Tuberous sclerosis
TTC19	Mitochondrial complex III deficiency, nuclear type 2
UBA5	Epileptic encephalopathy, early infantile, 44, Spinocerebellar ataxia, autosomal recessive 24
UBE3A	Angelman syndrome
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2
VARS	Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum (PEBAT), Encephalopathy, progressive
WARS2	Encephalopathy, mitochondrial
WDR45	Neurodegeneration with brain iron accumulation
WWOX	Epileptic encephalopathy, early infantile, Spinocerebellar ataxia
ZEB2	Mowat-Wilson syndrome