iGen
Tegevusluba L02423

Leukodüstroofia ja leukotsefalopaatia NGS paneel

Üldine info

Analüüsi kirjeldus: Leukodüstroofia ja leukotsefalopaatiaga seotud geenide uurimine. Uuritakse 116 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ABCD1, ADAR, AIFM1, AIMP1, ALDH3A2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARSA, ASPA, CLCN2, COA7, COL4A1, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS, DARS2, DEGS1, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS, FA2H, FAM126A, FDX1L, FOLR1, FOXRED1, GALC, GFAP, GFM1, GJC2, HEPACAM, HIBCH, HSPD1, HTRA1, IBA57, L2HGDH, LMNB1, LYRM7, MARS2, MLC1, 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-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, NDUFAF5, NFU1, NKX6-2, NOTCH3, NT5C2, NUBPL, PLP1, POLR3A, POLR3B, PSAP, PYCR2, RARS, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, SAMHD1, SCO1, SDHAF1, SERAC1, SLC1A4, SNORD118, SOX10, SUMF1, TREX1, TTC19, TUBB4A, ZFYVE26*

Haigekassa kood: 66618x3

Meetod: 116 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)**

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.

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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: Leukodüstroofiad ja leukoentsefalopaatiad on pärilikud ajuatroofiad, mis aju piltdiagnostikas avalduvad ajusiseste ning ajuväliste vedelikuruumide suurenemisega, võivad mõjutada ka perifeerset müeliniseerumist.
Leukodüstroofiad on päritavad autosoomdominantselt, -retsessiivselt või X-liiteliselt.. Leukoentsefalopaatia võib samuti olla sarnaselt pärilik ja põhjustades eelpoolkirjaldatule sarnaseid kõrvalekaldeid, kuid nende fenotüüp ei vasta leukodüstroofia kitsamatele kriteeriumidele.
Geneetiline testimine on näidustatud diagnoosi täpsustamiseks ja geneetiliseks nõustamiseks.
Vanderver A, et al.,; GLIA Consortium. Case definition and classification of leukodystrophies and leukoencephalopathies. Mol Genet Metab. 2015 Apr;114(4):494-500. doi: 10.1016/j.ymgme.2015.01.006. Epub 2015 Jan 29. PMID: 25649058; PMCID: PMC4390457.

Geenide nimekiri (116):

Geen	Fenotüüp
ABCD1	Adrenoleukodystrophy
ADAR	Dyschromatosis symmetrica hereditaria, Aicardi-Goutières syndrome
AIFM1	Deafness, Combined oxidative phosphorylation deficiency 6, Cowchock syndrome
AIMP1	Leukodystrophy, hypomyelinating
ALDH3A2	Sjogren-Larsson syndrome
AP4B1	Spastic paraplegia 47, autosomal recessive
AP4E1	Stuttering, familial persistent, 1, Spastic paraplegia 51, autosomal recessive
AP4M1	Spastic paraplegia 50, autosomal recessive
AP4S1	Spastic paraplegia 52, autosomal recessive
APOPT1	Mitochondrial complex IV deficiency
ARSA	Metachromatic leukodystrophy
ASPA	Aspartoacylase deficiency (Canavan disease)
CLCN2	Leukoencephalopathy with ataxia, Epilepsy
COA7	Spinocerebellar ataxia, Charcot-Marie-Tooth disease

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COL4A1	Schizencephaly, Anterior segment dysgenesis with cerebral involvement, Retinal artery tortuosity, Porencephaly, Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, Brain small vessel disease
COX15	Leigh syndrome, Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
COX6B1	Mitochondrial complex IV deficiency
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids
CTC1	Cerebroretinal microangiopathy with calcifications and cysts
CYP27A1	Cerebrotendinous xanthomatosis
D2HGDH	D-2-hydroxyglutaric aciduria 1
DARS	Hypomyelination with brainstem and spinal cord involvement and leg spasticity
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
DEGS1	Leukodystrophy, hypomyelinating
EARS2	Combined oxidative phosphorylation deficiency
EIF2B1	Leukoencephalopathy with vanishing white matter, Ovarioleukodystrophy
EIF2B2	Leukoencephalopathy with vanishing white matter, Ovarioleukodystrophy
EIF2B3	Leukoencephalopathy with vanishing white matter, Ovarioleukodystrophy
EIF2B4	Leukoencephalopathy with vanishing white matter, Ovarioleukodystrophy
EIF2B5	Leukoencephalopathy with vanishing white matter, Ovarioleukodystrophy
EPRS	Leukodystrophy, hypomyelinating
FA2H	Spastic paraplegia
FAM126A	Leukodystrophy, hypomyelinating
FDX1L	Myopathy
FOLR1	Cerebral folate deficiency
FOXRED1	Leigh syndrome, Mitochondrial complex I deficiency
GALC	Krabbe disease
GFAP	Alexander disease
GFM1	Combined oxidative phosphorylation deficiency
GJC2	Spastic paraplegia, Lymphedema, hereditary, Leukodystrophy, hypomyelinating
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts, remitting
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HSPD1	Spastic paraplegia, Leukodystrophy, hypomyelinating

HTRA1	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy type 2 (CADASIL2), Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL)
IBA57	Multiple mitochondrial dysfunctions syndrome 3, Spastic paraplegia 74, autosomal recessive
L2HGDH	L-2-hydroxyglutaric aciduria
LMNB1	Leukodystrophy, demyelinating, adult-onset, autosomal dominant
LYRM7	Mitochondrial complex III deficiency, nuclear type 8
MARS2	Combined oxidative phosphorylation deficiency
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
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-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
NDUFAF5	Mitochondrial complex I deficiency
NFU1	Multiple mitochondrial dysfunctions syndrome 1
NKX6-2	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy
NOTCH3	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL), Lateral meningocele syndrome
NT5C2	Spastic paraplegia 45
NUBPL	Mitochondrial complex I deficiency

PLP1	Spastic paraplegia, Pelizaeus-Merzbacher disease
POLR3A	Leukodystrophy, hypomyelinating
POLR3B	Leukodystrophy, hypomyelinating
PSAP	Krabbe disease, atypical, Metachromatic leukodystrophy due to saposin-b deficiency, Combined saposin deficiency, Gaucher disease, atypical, due to saposin C deficiency
PYCR2	Leukodystrophy, hypomyelinating 10
RARS	Leukodystrophy, hypomyelinating 9
RNASEH2A	Aicardi-Goutières syndrome
RNASEH2B	Aicardi-Goutières syndrome
RNASEH2C	Aicardi-Goutières syndrome
RNASET2	Leukoencephalopathy, cystic, without megalencephaly
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism (Gordon Holmes syndrome)
SAMHD1	Aicardi-Goutières syndrome, Chilblain lupus 2
SCO1	Mitochondrial complex IV deficiency
SDHAF1	Mitochondrial complex II deficiency
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly
SNORD118	Leukoencephalopathy, brain calcifications, and cysts (Labrune syndrome)
SOX10	Peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome, and Hirschsprung disease, Kallmann syndrome
SUMF1	Multiple sulfatase deficiency
TREX1	Vasculopathy, retinal, with cerebral leukodystrophy, Chilblain lupus, Aicardi-Goutières syndrome
TTC19	Mitochondrial complex III deficiency, nuclear type 2
TUBB4A	Leukodystrophy, hypomyelinating, Dystonia
ZFYVE26	Spastic paraplegia 15