iGen
Tegevusluba L02423

Lüsosomaalne ladestushaiguse NGS paneel

Üldine info

Analüüsi kirjeldus: Lüsosomaalne ladestushaigusega seotud geenide uurimine. Uuritakse 100 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ABCC8, ACY1, ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ANTXR2, ARG1, ARSA, ARSB, ASAH1, ASPA, ATP13A2, BTD, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSA, CTSC, CTSD, CTSK, DHCR7, DPYD, DYM, ETFA, ETFB, ETFDH, FH, FOLR1, FUCA1, GAA, GALC, GALNS, GAMT, GBA, GCDH, GLA, GLB1, GLDC, GM2A, GNE, GNPTAB, GNPTG, GNS, GPC3, GUSB, HEXA, HEXB, HGSNAT, HRAS, HYAL1, IDS*, IDUA, L2HGDH, LAMA2, LAMP2, LDB3, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, MOCS2, MYOT, NAGA, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX16, PEX26, PEX3, PEX5, PEX6, PGK1, PHYH, PPT1, PRODH, PSAP, QDPR, RAI1, SGSH, SLC17A5, SLC25A15, SLC46A1, SMPD1, SUMF1, SUOX, TCF4, TPP1*

Haigekassa kood: 66618x3

Meetod: 100 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.

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**.

iGen - Molekulaardiagnostika
Mäealuse 4/Akadeemia tee 15, 12618 Tallinn
Tel: 6000 199
Email: info@igen.ee www.igen.ee

iGen
Tegevusluba L02423

Teostamise aeg: kuni 4 nädalat

Geenide nimekiri (100):

Geen	Fenotüüp
ABCC8	Hyperinsulinemic hypoglycemia, Diabetes, permanent neonatal, Hypoglycemia, leucine-induced, Diabetes mellitus, transient neonatal, Pulmonary arterial hypertension (PAH)
ACY1	Aminoacylase 1 deficiency
ADSL	Adenylosuccinase deficiency
AGA	Aspartylglucosaminuria
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency
ALDH7A1	Epilepsy, pyridoxine-dependent
AMT	Glycine encephalopathy
ANTXR2	Hyalinosis, infantile systemic, Fibromatosis, juveline hyaline
ARG1	Hyperargininemia
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis (Maroteaux-Lamy)
ASAH1	Spinal muscular atrophy with progressive myoclonic epilepsy, Farber lipogranulomatosis
ASPA	Aspartoacylase deficiency (Canavan disease)
ATP13A2	Parkinson disease (Kufor-Rakeb syndrome)
BTD	Biotinidase deficiency
CLN3	Neuronal ceroid lipofuscinosis, type 3
CLN5	Neuronal ceroid lipofuscinosis, type 5
CLN6	Neuronal ceroid lipofuscinosis, type 6
CLN8	Neuronal ceroid lipofuscinosis, type 8
COL11A2	Weissenbacher-Zweymuller syndrome, Deafness, Otospondylomegaepiphyseal dysplasia, Fibrochondrogenesis, Stickler syndrome type 3 (non-ocular)
COL2A1	Avascular necrosis of femoral head, Rhegmatogenous retinal detachment, Epiphyseal dysplasia, with myopia and deafness, Czech dysplasia, Achondrogenesis type 2, Platyspondylic dysplasia Torrance type, Hypochondrogenesis, Spondyloepiphyseal dysplasia congenital (SEDC), Spondyloepimetaphyseal dysplasia (SEMD) Strudwick type, Kniest dysplasia, Spondyloperipheral dysplasia, Mild SED with premature onset arthrosis, SED with metatarsal shortening, Stickler syndrome type 1

iGen - Molekulaardiagnostika
Mäealuse 4/Akadeemia tee 15, 12618 Tallinn
Tel: 6000 199
Email: info@igen.ee www.igen.ee

CTNS	Cystinosis
CTSA	Galactosialidosis
CTSC	Periodontitis, juvenile, Haim-Munk syndrome, Papillon-Lefevre syndrome
CTSD	Ceroid lipofuscinosis, neuronal
CTSK	Pycnodysostosis
DHCR7	Smith-Lemli-Opitz syndrome
DPYD	5-fluorouracil toxicity, Developmental delay with or without dysmorphic facies and autism
DYM	Dyggve-Melchior-Clausen dysplasia, Smith-McCort dysplasia
ETFA	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
ETFB	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
ETFDH	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
FH	Hereditary leiomyomatosis and renal cell cancer
FOLR1	Cerebral folate deficiency
FUCA1	Fucosidosis
GAA	Glycogen storage disease
GALC	Krabbe disease
GALNS	Mucopolysaccharidosis (Morquio syndrome)
GAMT	Guanidinoacetate methyltransferase deficiency
GBA	Gaucher disease
GCDH	Glutaric aciduria
GLA	Fabry disease
GLB1	GM1-gangliosidosis, Mucopolysaccharidosis (Morquio syndrome)
GLDC	Glycine encephalopathy
GM2A	GM2-gangliosidosis, AB variant
GNE	Proximal myopathy and ophthalmoplegia, Nonaka myopathy, Sialuria
GNPTAB	Mucolipidosis
GNPTG	Mucolipidosis
GNS	Mucopolysaccharidosis (Sanfilippo syndrome)
GPC3	Simpson-Golabi-Behmel syndrome
GUSB	Mucopolysaccharidosis
HEXA	Tay-Sachs disease, GM2-gangliosidosis, Hexosaminidase A deficiency
HEXB	Sandhoff disease
HGSNAT	Mucopolysaccharidosis (Sanfilippo syndrome), Retinitis pigmentosa
HRAS	Costello syndrome, Congenital myopathy with excess of muscle spindles
HYAL1	Mucopolysaccharidosis

IDS	Mucopolysaccharidosis
IDUA	Mucopolysaccharidosis
L2HGDH	L-2-hydroxyglutaric aciduria
LAMA2	Muscular dystrophy, congenital merosin-deficient
LAMP2	Danon disease
LDB3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar
LIPA	Wolman disease, Cholesterol ester storage disease
MAN1B1	Mental retardation
MAN2B1	Mannosidosis, alpha B, lysosomal
MANBA	Mannosidosis, lysosomal
MCOLN1	Mucolipidosis
MFSD8	Ceroid lipofuscinosis, neuronal
MOCS2	Molybdenum cofactor deficiency
MYOT	Myopathy, myofibrillar, Muscular dystrophy, limb-girdle, 1A, Myopathy, spheroid body
NAGA	Kanzaki disease, Alpha-n-acetylgalactosaminidase deficiency, Schindler disease type I, Schindler disease type III
NAGLU	Mucopolysaccharidosis (Sanfilippo syndrome), Charcot-Marie-Tooth disease, axonal, type 2V
NEU1	Sialidosis
NPC1	Niemann-Pick disease
NPC2	Niemann-pick disease
PEX1	Heimler syndrome, Peroxisome biogenesis factor disorder 1A, Peroxisome biogenesis factor disorder 1B
PEX10	Adrenoleukodystrophy, neonatal, Zellweger syndrome, Peroxisome biogenesis disorder, Ataxia
PEX12	Zellweger syndrome, Peroxisome biogenesis disorder
PEX13	Adrenoleukodystrophy, neonatal, Zellweger syndrome, Peroxisome biogenesis disorder
PEX16	Zellweger syndrome, Peroxisome biogenesis disorder
PEX26	Adrenoleukodystrophy, neonatal, Zellweger syndrome, Peroxisome biogenesis disorder
PEX3	Zellweger syndrome, Peroxisome biogenesis disorder
PEX5	Adrenoleukodystrophy, neonatal, Rhizomelic chondrodysplasia punctata, Zellweger syndrome, Peroxisome biogenesis disorder
PEX6	Heimler syndrome, Peroxisome biogenesis disorder 4A, Peroxisome biogenesis disorder 4B
PGK1	Phosphoglycerate kinase 1 deficiency

PHYH	Refsum disease
PPT1	Ceroid lipofuscinosis, neuronal
PRODH	Hyperprolinemia
PSAP	Krabbe disease, atypical, Metachromatic leukodystrophy due to saposin-b deficiency, Combined saposin deficiency, Gaucher disease, atypical, due to saposin C deficiency
QDPR	Hyperphenylalaninemia, BH4-deficient
RAI1	Smith-Magenis syndrome
SGSH	Mucopolysaccharidosis (Sanfilippo syndrome)
SLC17A5	Sialuria, Finnish (Salla disease), Infantile sialic acid storage disorder
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC46A1	Folate malabsorption
SMPD1	Niemann-Pick disease
SUMF1	Multiple sulfatase deficiency
SUOX	Sulfocysteinuria
TCF4	Corneal dystrophy, Fuchs endothelial, Pitt-Hopkins syndrome
TPP1	Spinocerebellar ataxia, Neuronal ceroid lipofuscinosis type 2