Tegevusluba L02423

Metaboolne müopaatia ja rabdomüolüüsi NGS paneel

Üldine info

Analüüsi kirjeldus: Metaboolse müopaatia ja rabdomüolüüsiga seotud geenide uurimine. Uuritakse 88 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ACAD9, ACADL, ACADM, ACADVL, ADCK3, AGL, ALDOA, AMPD1, ANO5, C10ORF2, CAV3, COQ2, CPT2, DYSF, ENO3, ETFA, ETFB, ETFDH, FKRP, FKTN, FLAD1, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, 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-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, MYH3, OPA1, OPA3, PFKM, PGAM2, PGK1, PGM1, PHKA1, POLG, POLG2, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SLC22A5, SLC25A20, SUCLA2, SUCLG1, TANGO2, TK2, TYMP*

Haigekassa kood: 66618x3

Meetod: 88 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**.

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Teostamise aeg: kuni 4 nädalat

Geenide nimekiri (88):

Geen	Fenotüüp
ACAD9	Acyl-CoA dehydrogenase family, deficiency
ACADL	Long chain acyl-CoA dehydrogenase deficiency
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency
ACADVL	Acyl-CoA dehydrogenase, very long chain, deficiency
ADCK3	Coenzyme Q10 deficiency, Progressive cerebellar ataxia and atrophy, Spinocerebellar ataxia
AGL	Glycogen storage disease
AHCY	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency
ALDOA	Glycogen storage disease
AMPD1	Myoadenylate deaminase deficiency
ANO5	Gnathodiaphyseal dysplasia, LGMD2L and distal MMD3 muscular dystrophies
C10ORF2	Perrault syndrome, Mitochondrial DNA depletion syndrome, Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3
CAV3	Creatine phosphokinase, elevated serum, Hypertrophic cardiomyopathy (HCM), Long QT syndrome, Muscular dystrophy, limb-girdle, type IC, Myopathy, distal, Tateyama type, Rippling muscle disease 2
COQ2	Coenzyme Q10 deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
DYSF	Miyoshi muscular dystrophy, Muscular dystrophy, limb-girdle, Myopathy, distal, with anterior tibial onset
ENO3	Glycogen storage disease
ETFA	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
ETFB	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
ETFDH	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
FKRP	Muscular dystrophy-dystroglycanopathy

FKTN	Muscular dystrophy-dystroglycanopathy, Dilated cardiomyopathy (DCM), Muscular dystrophy-dystroglycanopathy (limb-girdle)
FLAD1	Lipid storage myopathy due to FLAD1 deficiency (LSMFLAD)
GAA	Glycogen storage disease
GBE1	Glycogen storage disease
GYG1	Glycogen storage disease, Polyglucosan body myopathy 2
GYS1	Glycogen storage disease
HADHA	Trifunctional protein deficiency, Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HADHB	Trifunctional protein deficiency
ISCU	Myopathy with lactic acidosis
LDHA	Glycogen storage disease
LPIN1	Myoglobinuria, acute, recurrent
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-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
MYH3	Arthrogryposis
OPA1	Optic atrophy, Optic atrophy 1, Optic atrophy with or without deafness, Ophthalmoplegia, myopathy, ataxia, and neuropathy, Behr synrome, Mitochondrial DNA depletion syndrome 14
OPA3	Optic atrophy, 3-methylglutaconic aciduria
PFKM	Glycogen storage disease
PGAM2	Glycogen storage disease
PGK1	Phosphoglycerate kinase 1 deficiency
PGM1	Congenital disorder of glycosylation
PHKA1	Glycogen storage disease
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
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions
PYGM	Glycogen storage disease
RBCK1	Polyglucosan body myopathy
RRM2B	Progressive external ophthalmoplegia with mitochondrial DNA deletions, Mitochondrial DNA depletion syndrome
RYR1	Central core disease, Malignant hyperthermia, Minicore myopathy with external ophthalmoplegia, Centronuclear myopathy, Minicore myopathy, Multicore myopathy
SCN4A	Hyperkalemic periodic paralysis, Myotonia, potassium-aggravated, Paramyotonia congenita, Myasthenic syndrome, congenital, Normokalemic potassium-sensitive periodic paralysis
SLC22A5	Carnitine deficiency, systemic primary
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SUCLA2	Mitochondrial DNA depletion syndrome
SUCLG1	Mitochondrial DNA depletion syndrome
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration (MECRCN)

TK2	Mitochondrial DNA depletion syndrome
TYMP	Mitochondrial DNA depletion syndrome

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