iGen
Tegevusluba L02423

Kardiomüopaatia NGS paneel

Üldine info

Analüüsi kirjeldus: Kardiomüopaatiaga seotud geenide uurimine. Uuritakse 214 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *AARS2, ABCC6, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALMS1, ALPK3, ANO5, APOA1, BAG3, BRAF, CALR3, CAPN3, CASQ2, CASZ1, CBL, CDH2, CHRM2, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DBH, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, ELAC2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXL4, FBXO32, FHL1, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GATC, GBE1, GFM1, GLA, GLB1, GMPPB, GSK3B, GTPBP3, GUSB, HADHA, HAND1, HCN4, HFE, HRAS, IDUA, ILK, ISPD, JPH2, JUP, KLHL24, KRAS, LAMA2, LAMP2, LARGE, LDB3, LEMD2, LMNA, LMOD2, LRRC10, LZTR1, MAP2K1, MAP2K2, MAP3K8, MIPEP, MLYCD, 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, MTO1, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYOT, MYPN, MYRF, NDUFAF2, NEXN, NF1, NKX2-5, NONO, NRAP, NRAS, PCCA, PCCB, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, PPA2, PPCS, PPP1CB, PRDM16, PRKAG2, PTPN11, QRSL1, RAF1, RASA2, RBCK1, RBM20, RIT1, RMND1, RRAS, RYR2, SCN5A, SCNN1B, SCNN1G, SCO1, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A4, SMCHD1, SOS1, SOS2, SPEG, SPRED1, TAB2, TAZ, TBX20, TBX5, TCAP, TGFB3, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRIM32, TSFM, TTN, TTR, VCL, VCP, VPS13A, XK*

Haigekassa kood: 66618x4

Meetod: 214 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

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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: Kardiomüopaatiad on tugeva geneetilise taustaga raskete südamehaiguste rühm, mis on seotud südamepuudulikkuse ja südame äkksurma sündroomi märkimisväärselt suurenenud riskiga. Kardiomüopaatia fenotüübiga seotud mutatsioone on tänaseks iseloomustatud enam kui 100 geenist (geenid, mis kodeerivad nt sarkomeeri, tsütoskeleti, desmosoomi, ioonkanalite või tuumalaamina struktuuri valke ja valke, mis osalevad ioonkanalite töös.

Testimise näidustusteks on kliinilise diagnoosi kinnitamine, diferentsiaaldiagnostika, ja/või vajadus geneetiliseks konsultatsiooniks

Geenide nimekiri (214):

Geen	Fenotüüp
AARS2	Leukoencephalopathy, progressive, with ovarian failure, Combined oxidative phosphorylation deficiency 8
ABCC6	Pseudoxanthoma elasticum
ABCC9	Atrial fibrillation, Cantu syndrome, Dilated cardiomyopathy (DCM)
ACAD9	Acyl-CoA dehydrogenase family, deficiency
ACADVL	Acyl-CoA dehydrogenase, very long chain, deficiency
ACTA1	Myopathy

ACTC1	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Atrial septal defect, Dilated cardiomyopathy (DCM)
ACTN2	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
AGK	Sengers syndrome, Cataract 38
AGL	Glycogen storage disease
ALMS1	Alström syndrome
ALPK3	Pediatric cardiomyopathy
ANO5	Gnathodiaphyseal dysplasia, LGMD2L and distal MMD3 muscular dystrophies
APOA1	Amyloidosis, systemic nonneuronopathic, Hypoalphalipoproteinemia
BAG3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar
BRAF	LEOPARD syndrome, Noonan syndrome, Cardiofaciocutaneous syndrome
CALR3	Cardiomyopathy, familial hypertrophic, 19
CAPN3	Muscular dystrophy, limb-girdle, Eosinophilic myositis
CASQ2	Ventricular tachycardia, catecholaminergic, polymorphic
CASZ1	Dilated cardiomyopathy (DCM), Ventricular septal defect
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
CDH2	Arrhythmogenic right ventricular cardiomyopathy (ARVC), Neurodevelopmental disorder
CHRM2	Dilated cardiomyopathy (DCM)
COX15	Leigh syndrome, Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CRYAB	Cataract, myofibrillar myopathy and cardiomyopathy, Congenital cataract and cardiomyopathy, Dilated cardiomyopathy (DCM), Myopathy, myofibrillar, Cataract 16, multiple types, Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related
CSRP3	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
CTNNA3	Arrhythmogenic right ventricular dysplasia

DBH	Dopamine beta-hydroxylase deficiency
DES	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar, Scapuloperoneal syndrome, neurogenic, Kaeser type
DMD	Becker muscular dystrophy, Duchenne muscular dystrophy, Dilated cardiomyopathy (DCM)
DNAJC19	3-methylglutaconic aciduria
DOLK	Congenital disorder of glycosylation
DPM3	Congenital disorder of glycosylation, Dilated cardiomyopathy (DCM), Limb-girdle muscular dystrophy
DSC2	Arrhythmogenic right ventricular dysplasia with palmoplantar keratoderma and woolly hair, Arrhythmogenic right ventricular dysplasia
DSG2	Arrhythmogenic right ventricular dysplasia, Dilated cardiomyopathy (DCM)
DSP	Cardiomyopathy, dilated, with wooly hair, keratoderma, and tooth agenesis, Arrhythmogenic right ventricular dysplasia, familial, Cardiomyopathy, dilated, with wooly hair and keratoderma, Keratosis palmoplantaris striata II, Epidermolysis bullosa, lethal acantholytic
DTNA	Left ventricular noncompaction 1
DYSF	Miyoshi muscular dystrophy, Muscular dystrophy, limb-girdle, Myopathy, distal, with anterior tibial onset
EEF1A2	Epileptic encephalopathy, early infantile, Mental retardation
ELAC2	Combined oxidative phosphorylation deficiency 17
EMD	Emery-Dreifuss muscular dystrophy
EPG5	Vici syndrome
ETFA	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
ETFB	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
ETFDH	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency
FBXL4	Mitochondrial DNA depletion syndrome
FBXO32	Dilated cardiomyopathy (DCM)

FHL1	Myopathy with postural muscle atrophy, Emery-Dreifuss muscular dystrophy, Reducing bod myopathy
FHOD3	Cardiomyopathy, familial hypertrophic
FKRP	Muscular dystrophy-dystroglycanopathy
FKTN	Muscular dystrophy-dystroglycanopathy, Dilated cardiomyopathy (DCM), Muscular dystrophy-dystroglycanopathy (limb-girdle)
FLNC	Myopathy
FOXD4	Dilated cardiomyopathy (DCM)
FOXRED1	Leigh syndrome, Mitochondrial complex I deficiency
FXN	Friedreich ataxia
GAA	Glycogen storage disease
GATA4	Tetralogy of Fallot, Atrioventricular septal defect, Testicular anomalies with or without congenital heart disease, Ventricular septal defect, Atrial septal defect
GATA6	Heart defects, congenital, and other congenital anomalies, Atrial septal defect 9, atrioventricular septal defect 5, Persistent truncus arteriosus, Tetralogy of Fallot
GATAD1	Dilated cardiomyopathy (DCM)
GATC	Cardiomyopathy, fatal
GBE1	Glycogen storage disease
GFM1	Combined oxidative phosphorylation deficiency
GLA	Fabry disease
GLB1	GM1-gangliosidosis, Mucopolysaccharidosis (Morquio syndrome)
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), Limb-girdle muscular dystrophy-dystroglycanopathy
GSK3B	Hypertrophic cardiomyopathy, Dilated cardiomyopathy (DCM)
GTPBP3	Combined oxidative phosphorylation deficiency 23
GUSB	Mucopolysaccharidosis
HADHA	Trifunctional protein deficiency, Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HAND1	Congenital heart defects, Dilated cardiomyopathy

HCN4	Sick sinus syndrome, Brugada syndrome, Left ventricular non-compaction cardiomyopathy (LVNC)
HFE	Hemochromatosis
HRAS	Costello syndrome, Congenital myopathy with excess of muscle spindles
IDUA	Mucopolysaccharidosis
ILK	Dilated cardiomyopathy (DCM)
ISPD	Muscular dystrophy-dystroglycanopathy
JPH2	Hypertrophic cardiomyopathy (HCM)
JUP	Arrhythmogenic right ventricular dysplasia, Naxos disease
KLHL24	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, Dilated cardiomyopathy (DCM), Hypertrophic cardiomyopathy (HCM)
KRAS	Noonan syndrome, Cardiofaciocutaneous syndrome
LAMA2	Muscular dystrophy, congenital merosin-deficient
LAMP2	Danon disease
LARGE	Muscular dystrophy-dystroglycanopathy
LDB3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar
LEMD2	Cataract 46, juvenile onset, Arrhythmogenic right ventricular cardiomyopathy (ARVC), Dilated cardiomyopathy (DCM)
LMNA	Heart-hand syndrome, Slovenian, Limb-girdle muscular dystrophy, Muscular dystrophy, congenital, LMNA-related, Lipodystrophy (Dunnigan), Emery-Dreiffus muscular dystrophy, Malouf syndrome, Dilated cardiomyopathy (DCM), Mandibuloacral dysplasia type A, Progeria Hutchinson-Gilford type
LMOD2	Familial dilated cardiomyopathy
LRRC10	Dilated cardiomyopathy (DCM)
LZTR1	Schwannomatosis, Noonan syndrome
MAP2K1	Cardiofaciocutaneous syndrome
MAP2K2	Cardiofaciocutaneous syndrome
MAP3K8	Noonan syndrome
MIPEP	Combined oxidative phosphorylation deficiency 31

MLYCD	Malonyl-CoA decarboxylase deficiency
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
MTO1	Combined oxidative phosphorylation deficiency
MYBPC3	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)

MYBPHL	Dilated cardiomyopathy (DCM)
MYH6	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM), Atrial septal defect 3
MYH7	Hypertrophic cardiomyopathy (HCM), Myopathy, myosin storage, Myopathy, distal, Dilated cardiomyopathy (DCM)
MYL2	Hypertrophic cardiomyopathy (HCM), Infantile type I muscle fibre disease and cardiomyopathy
MYL3	Hypertrophic cardiomyopathy (HCM)
MYL4	Atrial fibrillation, familial, 18
MYOT	Myopathy, myofibrillar, Muscular dystrophy, limb-girdle, 1A, Myopathy, spheroid body
MYPN	Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Dilated cardiomyopathy (DCM), Nemaline myopathy 11, autosomal recessive
MYRF	Congenital heart malformations, Congenital abnormalities of the kidney and urinary tract
NDUFAF2	Mitochondrial complex I deficiency, Leigh syndrome
NEXN	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
NF1	Watson syndrome, Neurofibromatosis, Neurofibromatosis-Noonan syndrome
NKX2-5	Conotruncal heart malformations, Hypothyroidism, congenital nongoitrous,, Atrial septal defect, Ventricular septal defect 3, Conotruncal heart malformations, variable, Tetralogy of Fallot
NONO	Mental retardation, X-linked, syndrome 34, Left ventricular non-compaction cardiomyopathy (LVNC)
NRAP	Dilated cardiomyopathy (DCM)
NRAS	Noonan syndrome
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PKP2	Arrhythmogenic right ventricular dysplasia
PLEC	Muscular dystrophy, limb-girdle, Epidermolysis bullosa
PLEKHM2	Dilated cardiomyopathy (DCM), left ventricular noncompaction
PLN	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)

PNPLA2	Neutral lipid storage disease with myopathy
PPA2	Sudden cardiac failure, infantile
PPCS	Dilated cardiomyopathy (DCM)
PPP1CB	Noonan syndrome-like disorder with loose anagen hair 2
PRDM16	Left ventricular noncompaction, Dilated cardiomyopathy (DCM)
PRKAG2	Hypertrophic cardiomyopathy (HCM), Wolff-Parkinson-White syndrome, Glycogen storage disease of heart, lethal congenital
PTPN11	Noonan syndrome, Metachondromatosis
QRSL1	Mitochondrial multisystemic disorder
RAF1	LEOPARD syndrome, Noonan syndrome, Dilated cardiomyopathy (DCM)
RASA2	Noonan syndrome
RBCK1	Polyglucosan body myopathy
RBM20	Dilated cardiomyopathy (DCM)
RIT1	Noonan syndrome
RMND1	Combined oxidative phosphorylation deficiency
RRAS	Noonan-syndrome like phenotype
RYR2	Ventricular tachycardia, catecholaminergic polymorphic, Arrhythmogenic right ventricular dysplasia
SCN5A	Heart block, nonprogressive, Heart block, progressive, Long QT syndrome, Ventricular fibrillation, Atrial fibrillation, Sick sinus syndrome, Brugada syndrome, Dilated cardiomyopathy (DCM)
SCNN1B	Liddle syndrome, Pseudohypoaldosteronism, Bronchiectasis with or without elevated sweat chloride
SCNN1G	Liddle syndrome, Pseudohypoaldosteronism, Bronchiectasis with or without elevated sweat chloride
SCO1	Mitochondrial complex IV deficiency
SCO2	Leigh syndrome, Hypertrophic cardiomyopathy (HCM), Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, Myopia
SDHA	Leigh syndrome/Mitochondrial respiratory chain complex II deficiency, Gastrointestinal stromal tumor, Paragangliomas, Dilated cardiomyopathy (DCM), Cardiomyopathy, dilated, 1GG

SELENON	Muscular dystrophy, rigid spine, Myopathy, congenital, with fiber- disproportion
SGCA	Muscular dystrophy, limb-girdle
SGCB	Muscular dystrophy, limb-girdle
SGCD	Muscular dystrophy, limb-girdle, Dilated cardiomyopathy (DCM)
SGCG	Muscular dystrophy, limb-girdle
SHOC2	Noonan-like syndrome with loose anagen hair
SLC22A5	Carnitine deficiency, systemic primary
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions, Mitochondrial DNA depletion syndrome
SMCHD1	Facioscapulohumeral muscular dystrophy, Facioscapulohumeral muscular dystrophy, type 2
SOS1	Noonan syndrome
SOS2	Noonan syndrome 9
SPEG	Centronuclear myopathy 5
SPRED1	Legius syndrome
TAB2	Congenital heart defects, multiple types, 2
TAZ	3-Methylglutaconic aciduria, (Barth syndrome)
TBX20	Atrial septal defect 4
TBX5	Holt-Oram syndrome
TCAP	Muscular dystrophy, limb-girdle, Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
TGFB3	Loeys-Dietz syndrome (Reinhoff syndrome), Arrhythmogenic right ventricular dysplasia
TMEM43	Arrhythmogenic right ventricular dysplasia, Emery-Dreifuss muscular dystrophy
TMEM70	Mitochondrial complex V (ATP synthase) deficiency
TNNC1	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
TNNI3	Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Dilated cardiomyopathy (DCM)
TNNI3K	Cardiac conduction disease with or without dilated cardiomyopathy

TNNT2	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Dilated cardiomyopathy (DCM)
TOR1AIP1	Muscular dystrophy with progressive weakness, distal contractures and rigid spine
TPM1	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
TRIM32	Bardet-Biedl syndrome, Muscular dystrophy, limb-girdle
TSFM	Combined oxidative phosphorylation deficiency
TTN	Dilated cardiomyopathy (DCM), Tibial muscular dystrophy, Limb-girdle muscular dystrophy, Hereditary myopathy with early respiratory failure, Myopathy, early-onset, with fatal cardiomyopathy (Salih myopathy), Muscular dystrophy, limb-girdle, type 2J
TTR	Dystransthyretinemic hyperthyroxinemia, Amyloidosis, hereditary, transthyretin-related
VCL	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
VCP	Amyotrophic lateral sclerosis, Inclusion body myopathy with early-onset Paget disease, Charcot-Marie-Tooth disease
VPS13A	Choreoacanthocytosis
XK	McLeod syndrome