iGen
Tegevusluba L02423

Dilateeruv kardiomüopaatia NGS paneel

Üldine info

Analüüsi kirjeldus: Dilateeruv kardiomüopaatiaga seotud geenide uurimine. Uuritakse 123 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ABCC6, ABCC9, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, APOA1, BAG3, CASZ1, CHRM2, DES, DMD, DOLK, DPM3, DSC2, DSG2, DSP, DYSF, EEF1A2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXO32, FHOD3, FKTN, FLNC, FOXD4, GATA4, GATA6, GATC, GBE1, GLB1, GSK3B, HAND1, HCN4, ILK, JPH2, JUP, KLHL24, LAMP2, LDB3, LEMD2, LMNA, LMOD2, LRRC10, 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, MYBPC3, MYBPHL, MYH6, MYH7, MYL4, NKX2-5, NRAP, PCCA, PCCB, PKP2, PLEKHM2, PLN, PPCS, PRDM16, QRSL1, RAF1, RBCK1, RBM20, RMND1, SCN5A, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TTN, TTR, VCL, VPS13A*

Haigekassa kood: 66618x3

Meetod: 123 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: Dilateeruv kardiomüopaatia (DKM) on kardiomüopaatiate üks peamisi alamtüüpe. Seda seostatakse tavaliselt südamekambrite, eriti vasaku vatsakese olulise laienemisega, müokardi seina hõrenemisega ja süstoolsete düsfunktsioonidega. See on südamepuudulikkuse, südame äkksurma ja südame siirdamise kõige levinum põhjus. Viimase kümne aasta jooksul on DKM molekulaargeneetika mõistmisel toimunud suured mutused. Sarnaselt hüpertroofilise kardiomüopaatiaga on geeniuuringud muutumas standardiks mitte-isheemilise DKM patsientide diagnostikas. Suurte patsientide ja perekondade kohortide uuringud on näidanud, et DKM sagedus võib ulatuda 1: 250
Nooremas eas võib DKM kulgeda aastaid asümptomaatilisena, sümptomaatiliseks kujuneb see haiguse hilisemates järkudes, avaldudes rütmihäirete, südamepuudulikkuse ja trombemboolilise haigusena.
Testimise näidustusteks on kliinilise diagnoosi kinnitamine, diferentsiaaldiagnostika ja/või vajadus geneetiliseks konsultatsiooniks.

Geenide nimekiri (123):

Geen	Fenotüüp
ABCC6	Pseudoxanthoma elasticum
ABCC9	Atrial fibrillation, Cantu syndrome, Dilated cardiomyopathy (DCM)
ACTA1	Myopathy
ACTC1	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Atrial septal defect, Dilated cardiomyopathy (DCM)
ACTN2	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
ALMS1	Alström syndrome
ALPK3	Pediatric cardiomyopathy
APOA1	Amyloidosis, systemic nonneuronopathic, Hypoalphalipoproteinemia
BAG3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar
CASZ1	Dilated cardiomyopathy (DCM), Ventricular septal defect
CHRM2	Dilated cardiomyopathy (DCM)

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DES	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar, Scapuloperoneal syndrome, neurogenic, Kaeser type
DMD	Becker muscular dystrophy, Duchenne muscular dystrophy, Dilated cardiomyopathy (DCM)
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
DYSF	Miyoshi muscular dystrophy, Muscular dystrophy, limb-girdle, Myopathy, distal, with anterior tibial onset
EEF1A2	Epileptic encephalopathy, early infantile, Mental retardation
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
FBXO32	Dilated cardiomyopathy (DCM)
FHOD3	Cardiomyopathy, familial hypertrophic
FKTN	Muscular dystrophy-dystroglycanopathy, Dilated cardiomyopathy (DCM), Muscular dystrophy-dystroglycanopathy (limb-girdle)
FLNC	Myopathy
FOXD4	Dilated cardiomyopathy (DCM)
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
GATC	Cardiomyopathy, fatal
GBE1	Glycogen storage disease
GLB1	GM1-gangliosidosis, Mucopolysaccharidosis (Morquio syndrome)
GSK3B	Hypertrophic cardiomyopathy, Dilated cardiomyopathy (DCM)
HAND1	Congenital heart defects, Dilated cardiomyopathy
HCN4	Sick sinus syndrome, Brugada syndrome, Left ventricular non-compaction cardiomyopathy (LVNC)

ILK	Dilated cardiomyopathy (DCM)
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)
LAMP2	Danon disease
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)
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
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)
MYL4	Atrial fibrillation, familial, 18
NKX2-5	Conotruncal heart malformations, Hypothyroidism, congenital nongoitrous,, Atrial septal defect, Ventricular septal defect 3, Conotruncal heart malformations, variable, Tetralogy of Fallot
NRAP	Dilated cardiomyopathy (DCM)
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PKP2	Arrhythmogenic right ventricular dysplasia
PLEKHM2	Dilated cardiomyopathy (DCM), left ventricular noncompaction
PLN	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
PPCS	Dilated cardiomyopathy (DCM)
PRDM16	Left ventricular noncompaction, Dilated cardiomyopathy (DCM)
QRSL1	Mitochondrial multisystemic disorder
RAF1	LEOPARD syndrome, Noonan syndrome, Dilated cardiomyopathy (DCM)
RBCK1	Polyglucosan body myopathy
RBM20	Dilated cardiomyopathy (DCM)
RMND1	Combined oxidative phosphorylation deficiency
SCN5A	Heart block, nonprogressive, Heart block, progressive, Long QT syndrome, Ventricular fibrillation, Atrial fibrillation, Sick sinus syndrome, Brugada syndrome, Dilated cardiomyopathy (DCM)
SPEG	Centronuclear myopathy 5
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)
TMEM43	Arrhythmogenic right ventricular dysplasia, Emery-Dreifuss muscular dystrophy
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)
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)
VPS13A	Choreoacanthocytosis