iGen
Tegevusluba L02423

Arütmia NGS paneel

Üldine info

Analüüsi kirjeldus: Arütmiaga seotud geenide uurimine. Uuritakse 58 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ABCC9, AKAP9, ANK2, BAG3, CACNA1C, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DBH, DES, DSC2, DSG2, DSP, FLNC, GATA6, HADHA, HCN4, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LEMD2, LMNA, MYH6, MYH7, MYL4, NKX2-5, NOS1AP, NUP155, PKP2, PLN, PPA2, RYR2, SALL4, SCN10A, SCN1B, SCN3B, SCN5A, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TNNI3K, TNNT2, TRDN, TRPM4, TTN*

Haigekassa kood: 66618x3

Meetod: 58 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**.

Teostamise aeg: kuni 4 nädalat

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Geenide nimekiri (58):

Geen	Fenotüüp
ABCC9	Atrial fibrillation, Cantu syndrome, Dilated cardiomyopathy (DCM)
AKAP9	Long QT syndrome
ANK2	Cardiac arrhythmia, Long QT syndrome
BAG3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar
CACNA1C	Brugada syndrome, Timothy syndrome
CACNB2	Brugada syndrome
CALM1	Ventricular tachycardia, catecholaminergic polymorphic, Recurrent cardiac arrest, infantile, Long QT syndrome
CALM2	Long QT syndrome
CALM3	Catecholaminergic polymorphic ventricular tachycardia
CASQ2	Ventricular tachycardia, catecholaminergic, polymorphic
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
CDH2	Arrhythmogenic right ventricular cardiomyopathy (ARVC), Neurodevelopmental disorder
CTNNA3	Arrhythmogenic right ventricular dysplasia
DBH	Dopamine beta-hydroxylase deficiency
DES	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar, Scapuloperoneal syndrome, neurogenic, Kaeser type
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
FLNC	Myopathy
GATA6	Heart defects, congenital, and other congenital anomalies, Atrial septal defect 9, atrioventricular septal defect 5, Persistent truncus arteriosus, Tetralogy of Fallot
HADHA	Trifunctional protein deficiency, Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HCN4	Sick sinus syndrome, Brugada syndrome, Left ventricular non-compaction cardiomyopathy (LVNC)

JUP	Arrhythmogenic right ventricular dysplasia, Naxos disease
KCNA5	Atrial fibrillation
KCNE1	Long QT syndrome, Jervell and Lange-Nielsen syndrome
KCNE2	Long QT syndrome, Atrial fibrillation, familial
KCNH2	Short QT syndrome, Long QT syndrome
KCNJ2	Short QT syndrome, Andersen syndrome, Long QT syndrome, Atrial fibrillation
KCNJ5	Long QT syndrome, Hyperaldosteronism, familial
KCNQ1	Short QT syndrome, Long QT syndrome, Atrial fibrillation, Jervell and Lange-Nielsen syndrome
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
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
NOS1AP	Romano-Ward syndrome
NUP155	Atrial fibrillation 15
PKP2	Arrhythmogenic right ventricular dysplasia
PLN	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)
PPA2	Sudden cardiac failure, infantile
RYR2	Ventricular tachycardia, catecholaminergic polymorphic, Arrhythmogenic right ventricular dysplasia
SALL4	Acro-renal-ocular syndrome, Duane-radial ray/Okohiro syndrome

SCN10A	Paroxysmal extreme pain disorder, Channelopathy-associated congenital insensitivity to pain, Primary erythermalgia, Sodium channelopathy-related small fiber neuropathy, Brugada syndrome
SCN1B	Atrial fibrillation, Brugada syndrome, Generalized epilepsy with febrile seizures plus, Epilepsy, generalized, with febrile seizures plus, type 1, Epileptic encephalopathy, early infantile, 52
SCN3B	Atrial fibrillation, familial, Brugada syndrome
SCN5A	Heart block, nonprogressive, Heart block, progressive, Long QT syndrome, Ventricular fibrillation, Atrial fibrillation, Sick sinus syndrome, Brugada syndrome, Dilated cardiomyopathy (DCM)
TBX5	Holt-Oram syndrome
TECRL	Ventricular tachycardia, catecholaminergic polymorphic, 3
TGFB3	Loeys-Dietz syndrome (Reinhoff syndrome), Arrhythmogenic right ventricular dysplasia
TMEM43	Arrhythmogenic right ventricular dysplasia, Emery-Dreifuss muscular dystrophy
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)
TRDN	Ventricular tachycardia, catecholaminergic polymorphic
TRPM4	Progressive familial heart block
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