iGen
Tegevusluba L02423

Parkinsoni NGS paneel

Üldine info

Analüüsi kirjeldus: Parkinsoniga seotud geenide uurimine. Uuritakse 60 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ATP13A2, ATP1A3, DNAJC6, FBXO7, GCH1, LRRK2, MAPT, 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-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, PARK2, PARK7, PDGFB, PDGFRB, PINK1, PLA2G6, PRKRA, SLC20A2, SLC39A14, SLC6A3, SNCA, SPR, SYNJ1, TH, VPS13A, VPS13C, VPS35*

Haigekassa kood: 66618x3

Meetod: 60 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 (60):

Geen	Fenotüüp
ATP13A2	Parkinson disease (Kufor-Rakeb syndrome)
ATP1A3	Alternating hemiplegia of childhood, Dystonia 12
DNAJC6	Juvenile Parkinsonism
FBXO7	Parkinson disease
GCH1	Dopa-Responsive Dystonia Hyperphenylalaninemia, BH4-deficient, GTP Cyclohydrolase 1-Deficient Dopa-Responsive Dystonia
LRRK2	Dementia, Lewy body, Parkinson disease
MAPT	Pick disease, Frontotemporal dementia, Parkinson-dementia syndrome, Supranuclear palsy, progressive
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

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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-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
PARK2	Parkinson disease, juvenile
PARK7	Parkinson disease, early onset
PDGFB	Basal ganglia calcification, idiopathic, 5
PDGFRB	Basal ganglia calcification, idiopathic, 4, Kosaki overgrowth syndrome, Premature aging syndrome, Penttinen type
PINK1	Parkinson disease, early onset
PLA2G6	Parkinson disease, Neurodegeneration with brain iron accumulation
PRKRA	Dystonia 16
SLC20A2	Basal ganglia calcification, idiopathic, 1
SLC39A14	Hypermanganesemia with dystonia 2
SLC6A3	Parkinsonism-dystonia, infantile
SNCA	Parkinson disease, Dementia with Lewy bodies
SPR	Dystonia, Dopa-responsive, due to sepiapterin reductase deficiency
SYNJ1	Epileptic encephalopathy, early infantile, 53, Parkinson disease 20, early-onset
TH	Segawa syndrome, autosomal recessive
VPS13A	Choreoacanthocytosis
VPS13C	Parkinson disease 23, autosomal recessive, early onset
VPS35	Parkinson disease