iGen
Tegevusluba L02423

Düstoonia NGS paneel

Üldine info

Analüüsi kirjeldus: Düstooniaga seotud geenide uurimine. Uuritakse 67 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ADCY5, ANO3, ATP1A3, BCAP31, CACNA1B, CACNA1G, DCAF17, DNAJC12, FA2H, FITM2, GCH1, GNAL, KCNMA1, KMT2B, MECR, MIPEP, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, M, 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, PDE10A, PDGFB, PDGFRB, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SLC39A14, SPR, TH, THAP1, TOR1A, UBTF*

Haigekassa kood: 66618x3

Meetod: 67 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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Näidustused:

Düstoonia on liikumishäire, mida iseloomustavad püsivad või vahelduvad lihaste tahtmatust kontraktsioonist tulenevad spasmid või poosid. Düstooniate ulatus on erinev, alates ühe lihase või lihasgrupi haaratusest kuni generaliseeritud düstooniateni. Ühel inimesel on enamasti tegemist stereotüüpse lihaste kontraktsiooniga. Osadel patsientidest esineb düstooniline treemor. Düstooniad jagunevad primaarseteks (idiopaatilisteks) ja sekundaarseteks (sümptomaatilisteks).
Paneelis uuritakse nii isoleeritud düstoonia pärilikke monogeenseid vorme kui ka kombineeritud düstooniaid/düskineesiaid (parkinsonismi, müokloonuselisi düstooniaid). Testimise näidustusteks on kliinilise diagnoosi kinnitamine ja diferentsiaaldiagnostika ja/või vajadus geneetiliseks konsultatsiooniks
Petrucci S, Valente EM. Genetic issues in the diagnosis of dystonias. Front Neurol. 2013 Apr 10;4:34. doi: 10.3389/fneur.2013.00034. PMID: 23596437; PMCID: PMC3622056.

Geenide nimekiri (67):

Geen	Fenotüüp
ADCY5	Dyskinesia, familial, with facial myokymia
ANO3	Dystonia 24
ATP1A3	Alternating hemiplegia of childhood, Dystonia 12
BCAP31	Deafness, dystonia, and cerebral hypomyelination
CACNA1B	Dystonia 23, Early infantile epileptic encephalopathy
CACNA1G	Spinocerebellar ataxia 42
DCAF17	Woodhouse-Sakati syndrome
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, Dystonia, Other hyperphenylalaninemias
FA2H	Spastic paraplegia
FITM2	Dystonia, Deafness
GCH1	Dopa-Responsive Dystonia Hyperphenylalaninemia, BH4-deficient, GTP Cyclohydrolase 1-Deficient Dopa-Responsive Dystonia
GNAL	Primary torsion dystonia

KCNMA1	Paroxysmal nonkinesigenic dyskinesia 3 with or without generalized epilepsy (PNKD3), Cerebellar atrophy, developmental delay, and seizures (CADEDS)
KMT2B	Dystonia 28, childhood-onset
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities (DYTOABG)
MIPEP*	Combined oxidative phosphorylation deficiency 31
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
PDE10A	Striatal degeneration, autosomal dominant 2, Infantile-onset dyskinesia
PDGFB	Basal ganglia calcification, idiopathic, 5

PDGFRB	Basal ganglia calcification, idiopathic, 4, Kosaki overgrowth syndrome, Premature aging syndrome, Penttinen type
PNKD	Paroxysmal non-kinesigenic dyskinesia
PRKRA	Dystonia 16
PRRT2	Episodic kinesigenic dyskinesia, Seizures, benign familial infantile, 2, Convulsions, familial infantile, with paroxysmal choreoathetosis
SGCE	Dystonia 11, myoclonic
SLC2A1	Stomatin-deficient cryohydrocytosis with neurologic defects, Epilepsy, idiopathic generalized, GLUT1 deficiency syndrome
SLC39A14	Hypermanganesemia with dystonia 2
SPR	Dystonia, Dopa-responsive, due to sepiapterin reductase deficiency
TH	Segawa syndrome, autosomal recessive
THAP1	Dystonia 6, torsion
TOR1A	Dystonia 1, torsion
UBTF	Neurodegeneration, childhood-onset, with brain atrophy