iGen
Tegevusluba L02423

Ataksia NGS paneel

Üldine info

Analüüsi kirjeldus: Ataksiaga seotud geenide uurimine. Uuritakse 208 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ABCB7, ABHD12, ACO2, ADCK3, ADPRHL2, AFG3L2, AGTPBP1, AHI1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP1A3, ATP2B3, ATP8A2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C10ORF2, C12ORF4, C5ORF42, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CLCN2, CLN5, CLPP, COA7, COASY, COX20, CSTB, CWF19L1, CYP27A1, CYP2U1, DHPS, DNAJC19, DNMT1, DOCK3, EBF3, EEF2, ELOVL4, ELOVL5, FA2H, FBXL4, FDXR, FGF14, FLVCR1, FMR1, FXN, GBA2, GFAP, GOSR2, GRID2, GRM1, GSS, HARS2, HIBCH, INPP5E, IRF2BPL, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, KIF7, LAMA1, LARS2, LMNB1, LRPPRC, MARS2, MECR, MKKS, MKS1, MME, MRE11A, MSTO1, 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, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NKX6-2, NOL3, NPHP1, NUBPL, OFD1, OPA1, OPHN1, PAX6, PDYN, PEX7, PHYH, PNKD, PNKP, PNPLA6, POLG, PRKCG, PRRT2, PUM1, RNF216, RUBCN, SACS, SCYL1, SERAC1, SETX, SIL1, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRIM32, TTBK2, TTC19, TTC8, TTPA, TUBB4A, UBA5, UBTF, UCHL1, VAMP1, VLDLR, WDPCP, WDR81, WFS1, WWOX, ZFYVE26, ZNF423*

Haigekassa kood: 66618x4

Meetod: 208 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)**

iGen - Molekulaardiagnostika
Mäealuse 4/Akadeemia tee 15, 12618 Tallinn
Tel: 6000 199
Email: info@igen.ee www.igen.ee

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

Näidustused: Ataksiad, (s.h. väikeaju, episoodilised ja spinotserebellaarsed ataksiad) on liigutuste koordinatsioonihäire, mida põhjustab tahtele alluvate lihaste koostöö puudumine. Ataksia korral esineb sageli väikeaju atroofia. Ataksia võib olla ka pärilik ja juba lapse- või noorukieas avalduv, kuna ajukahjustus süveneb järk järgult ajas, siis haigus on pideva süvenemise tendentsiga.
Friedrichi ataksia kantakse edasi autosoomretsessiivselt (avaldub noorelt 10-20 eluaastal), Pierre-Marie ataksia aga autosoom-dominantselt (avaldub tavaliselt hilisemas eas). Suurem osa spastilistest ataksiatest on pärilikud retsessiivselt.
Geneetiline testimine on näidustatud diferentsiaaldiagnostilistel eesmärkidel ja geneetiliseks konsultatsiooniks.

Geenide nimekiri (208):

Geen	Fenotüüp
ABCB7	Anemia, sideroblastic, and spinocerebellar ataxia
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract
ACO2	Optic atrophy, Infantile cerebellar-retinal degeneration
ADCK3	Coenzyme Q10 deficiency, Progressive cerebellar ataxia and atrophy, Spinocerebellar ataxia
ADPRHL2	Neurodegeneration, childhood-onset, with brain atrophy
AFG3L2	Spastic ataxia, Spinocerebellar ataxia

AGTPBP1	Neuropathy
AHI1	Joubert syndrome
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency
ANO10	Spinocerebellar ataxia
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
ARL13B	Joubert syndrome
ARL6	Bardet-Biedl syndrome, Retinitis pigmentosa
ATCAY	Ataxia, cerebellar, Cayman
ATM	Breast cancer, Ataxia-Telangiectasia
ATP1A3	Alternating hemiplegia of childhood, Dystonia 12
ATP2B3	Spinocerebellar ataxia, X-linked 1
ATP8A2	Dysequilibrium syndrome
BBS1	Bardet-Biedl syndrome
BBS10	Bardet-Biedl syndrome
BBS12	Bardet-Biedl syndrome
BBS2	Bardet-Biedl syndrome, Retinitis pigmentosa
BBS4	Bardet-Biedl syndrome
BBS5	Bardet-Biedl syndrome
BBS7	Bardet-Biedl syndrome
BBS9	Bardet-Biedl syndrome
BEAN1	Spinocerebellar ataxia
C10ORF2	Perrault syndrome, Mitochondrial DNA depletion syndrome, Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3
C12ORF4	Developmental delay and seizures with or without movement abnormalities (DEDSM)
C5ORF42	Orofaciodigital syndrome, Joubert syndrome
CA8	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome
CACNA1A	Migraine, familial hemiplegic, Episodic ataxia, Spinocerebellar ataxia 6, Epileptic encephalopathy, early infantile, 42
CACNA1G	Spinocerebellar ataxia 42
CACNB4	Episodic ataxia, Epilepsy, idiopathic generalized, susceptibility to, 9
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation
CAPN1	Spastic paraplegia 76, autosomal recessive
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, FG syndrome, Mental retardation
CC2D2A	COACH syndrome, Joubert syndrome, Meckel syndrome
CCDC88C	Spinocerebellar ataxia

CEP290	Bardet-Biedl syndrome, Leber congenital amaurosis, Joubert syndrome, Senior-Loken syndrome, Meckel syndrome
CEP41	Joubert syndrome
CLCN2	Leukoencephalopathy with ataxia, Epilepsy
CLN5	Neuronal ceroid lipofuscinosis, type 5
CLPP	Deafness
COA7	Spinocerebellar ataxia, Charcot-Marie-Tooth disease
COASY	Neurodegeneration with brain iron accumulation 6
COX20	Mitochondrial complex IV deficiency
CSTB	Epilepsy, progressive myoclonic
CWF19L1	Spinocerebellar ataxia
CYP27A1	Cerebrotendinous xanthomatosis
CYP2U1	Spastic paraplegia 56, autosomal recessive
DHPS	
DNAJC19	3-methylglutaconic aciduria
DNMT1	Neuropathy, hereditary sensory, Cerebellar ataxia, deafness, and narcolepsy
DOCK3	Ataxia
EBF3	Hypotonia, ataxia, and delayed development syndrome (HADDS)
EEF2	Spinocerebellar ataxia
ELOVL4	Stargardt disease, Icthyosis, spastic quadriplegia, and mental retardation, Spinocerebellar ataxia
ELOVL5	Spinocerebellar ataxia
FA2H	Spastic paraplegia
FBXL4	Mitochondrial DNA depletion syndrome
FDXR	Auditory neuropathy and optic atrophy
FGF14	Spinocerebellar ataxia
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa
FMR1	Premature ovarian failure, Fragile X syndrome, Fragile X tremor/ataxia syndrome
FXN	Friedreich ataxia
GBA2	Cerebellar ataxia with spasticity
GFAP	Alexander disease
GOSR2	Epilepsy, progessive myoclonic
GRID2	Spinocerebellar ataxia
GRM1	Spinocerebellar ataxia
GSS	Glutathione synthetase deficiency
HARS2	Perrault syndrome
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency

INPP5E	Joubert syndrome, Mental retardation, truncal obesity, retinal dystrophy, and micropenis (MORM syndrome)
IRF2BPL	Neurodevelopmental disorder with hypotonia, seizures, and absent language
ITM2B	Dementia, familial Danish, Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, Cerebral amyloid angiopathy
ITPR1	Spinocerebellar ataxia
KCNA1	Episodic ataxia/myokymia syndrome
KCNC3	Spinocerebellar ataxia
KCND3	Brugada syndrome, Spinocerebellar ataxia 19, Spinocerebellar ataxia 22
KCNJ10	Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SESAME syndrome), Pendred syndrome, Enlarged vestibular aqueduct
KIF1C	Spastic ataxia
KIF7	Acrocallosal syndrome, Hydrolethalus syndrome, Al-Gazali-Bakalinova syndrome, Joubert syndrome
LAMA1	Poretti-Boltshauser syndrome
LARS2	Perrault syndrome, Hydrops, lactic acidosis, and sideroblastic anemia (HLASA)
LMNB1	Leukodystrophy, demyelinating, adult-onset, autosomal dominant
LRPPRC	Leigh syndrome, French-Canadian type
MARS2	Combined oxidative phosphorylation deficiency
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities (DYTOABG)
MKKS	Bardet-Biedl syndrome, McKusick-Kaufman syndrome
MKS1	Bardet-Biedl syndrome, Meckel syndrome
MME	Spinocerebellar ataxia 43, Charcot-Marie-Tooth disease, axonal, type 2T
MRE11A	Ataxia-telangiectasia-like disorder-1
MSTO1	Myopathy, mitochondrial, and ataxia
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
MTFMT	Combined oxidative phosphorylation deficiency 15
MTPAP	Spastic ataxia
MTTP	Abetalipoproteinemia
NDUFAF6	Mitochondrial complex I deficiency, Leigh syndrome
NDUFS2	Mitochondrial complex I deficiency
NDUFS4	Mitochondrial complex I deficiency, Leigh syndrome
NDUFS7	Mitochondrial complex I deficiency, Leigh syndrome
NDUFS8	Mitochondrial complex I deficiency, Leigh syndrome
NDUFV1	Mitochondrial complex I deficiency
NKX6-2	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy
NOL3	Myoclonus, familial cortical
NPHP1	Nephronophthisis, Joubert syndrome, Senior-Loken syndrome
NUBPL	Mitochondrial complex I deficiency
OFD1	Simpson-Golabi-Behmel syndrome, Retinitis pigmentosa, Orofaciodigital syndrome, Joubert syndrome
OPA1	Optic atrophy, Optic atrophy 1, Optic atrophy with or without deafness, Ophthalmoplegia, myopathy, ataxia, and neuropathy, Behr synrome, Mitochondrial DNA depletion syndrome 14
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance
PAX6	Aniridia, cerebellar ataxia, and mental retardation (Gillespie syndrome), Keratitis, Coloboma, ocular, Cataract with late-onset corneal dystrophy, Morning glory disc anomaly, Foveal hypoplasia, Aniridia, Optic nerve hypoplasia, Peters anomaly
PDYN	Spinocerebellar ataxia

PEX7	Refsum disease, Rhizomelic CDP type 1
PHYH	Refsum disease
PNKD	Paroxysmal non-kinesigenic dyskinesia
PNKP	Epileptic encephalopathy, early infantile, Ataxia-oculomotor
PNPLA6	Laurence-Moon syndrome, Boucher-Neuhauser syndrome, Spastic paraplegia 39
POLG	POLG-related ataxia neuropathy spectrum disorders, Sensory ataxia, dysarthria, and ophthalmoparesis, Alpers syndrome, Progressive external ophthalmoplegia with mitochondrial DNA deletions, Mitochondrial DNA depletion syndrome
PRKCG	Spinocerebellar ataxia
PRRT2	Episodic kinesigenic dyskinesia, Seizures, benign familial infantile, 2, Convulsions, familial infantile, with paroxysmal choreoathetosis
PUM1	Ataxia
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism (Gordon Holmes syndrome)
RUBCN	Spinocerebellar ataxia
SACS	Spastic ataxia, Charlevoix-Saguenay
SCYL1	Spinocerebellar ataxia, autosomal recessive 21
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome
SETX	Ataxia with oculomotor apraxia, Amyotrophic lateral sclerosis, juvenile, Spinocerebellar ataxia
SIL1	Marinesco-Sjogren syndrome
SLC1A3	Episodic ataxia
SLC20A2	Basal ganglia calcification, idiopathic, 1
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB
SLC2A1	Stomatin-deficient cryohydrocytosis with neurologic defects, Epilepsy, idiopathic generalized, GLUT1 deficiency syndrome
SLC52A2	Brown-Vialetto-Van Laere syndrome
SLC9A1	Spinocerebellar ataxia, autosomal recessive 19 (Lichtenstein-Knorr syndrome)
SLC9A6	Mental retardation, syndromic, Christianson
SNX14	Spinocerebellar ataxia
SPG7	Spastic paraplegia
SPTBN2	Spinocerebellar ataxia
STUB1	Spinocerebellar ataxia
SYNE1	Spinocerebellar ataxia, autosomal recessive 8
SYT14	Spinocerebellar ataxia
TCTN1	Joubert syndrome
TCTN2	Joubert syndrome, Meckel syndrome

TCTN3	Orofaciodigital syndrome (Mohr-Majewski syndrome), Joubert syndrome
TDP1	Spinocerebellar ataxia, with axonal neuropathy
TGM6	Spinocerebellar ataxia
TMEM138	Joubert syndrome
TMEM216	Joubert syndrome, Meckel syndrome
TMEM231	Joubert syndrome, Meckel syndrome
TMEM237	Joubert syndrome
TMEM240	Spinocerebellar ataxia
TMEM67	Nephronophthisis, COACH syndrome, Joubert syndrome, Meckel syndrome
TPP1	Spinocerebellar ataxia, Neuronal ceroid lipofuscinosis type 2
TRIM32	Bardet-Biedl syndrome, Muscular dystrophy, limb-girdle
TTBK2	Spinocerebellar ataxia
TTC19	Mitochondrial complex III deficiency, nuclear type 2
TTC8	Bardet-Biedl syndrome, Retinitis pigmentosa
TTPA	Ataxia with isolated vitamin E deficiency
TUBB4A	Leukodystrophy, hypomyelinating, Dystonia
UBA5	Epileptic encephalopathy, early infantile, 44, Spinocerebellar ataxia, autosomal recessive 24
UBTF	Neurodegeneration, childhood-onset, with brain atrophy
UCHL1	Parkinson disease 5, autosomal dominant, Spastic paraplegia 79, autosomal recessive
VAMP1	Spastic ataxia
VLDLR	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome
WDPCP	Meckel-Gruber syndrome, modifier, Bardet-Biedl syndrome, Congenital heart defects, hamartomas of tongue, and polysyndactyly
WDR81	Dysequilibrium syndrome
WFS1	Wolfram syndrome, Deafness, Wolfram-like syndrome, autosomal dominant, Deafness, autosomal dominant 6/14/38, Cataract 41
WWOX	Epileptic encephalopathy, early infantile, Spinocerebellar ataxia
ZFYVE26	Spastic paraplegia 15
ZNF423	Nephronophthisis, Joubert syndrome

iGen - Molekulaardiagnostika
Mäealuse 4/Akadeemia tee 15, 12618 Tallinn
Tel: 6000 199
Email: info@igen.ee www.igen.ee