iGen
Tegevusluba L02423

Autismi spektri häired NGS paneel

Üldine info

Analüüsi kirjeldus: Autismi spektri häiretega seotud geenide uurimine. Uuritakse 75 geeni kõiki kodeerivaid eksoneid ja ekson-intron piirialasid

Geenid: *ADNP, BCL11A, C12ORF4, CACNA1C, CC2D1A, CNOT3, CNTN6, COL4A3BP, CSNK2A1, CTNND2, DHCR7, EHMT1, EN2, FBXO11, FOXP1, GAMT, KMT2E, KMT5B, MBOAT7, MECP2, 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, NBEA, NFIB, NLGN3, NLGN4X, NSD1, POGZ, PTCHD1, PTEN, RPL10, SHANK3, TBR1, TCF20, TRIP12, TSC1, TSC2, VAMP2, WASF1, ZSWIM6*

Haigekassa kood: 66618x3

Meetod: 75 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: Autismispektri häiret iseloomustavad sotsiaalse suhtluse puudujäägid, keele arengu puudumine või hilinemine, stereotüüpne korduvkäitumine ja jäigad käitumismustrid. Autismil on erinevaid etioloogiaid, kuna seda on kirjeldatud paljude neuroloogilise sündroomide korral. Autismi geneetiline põhjus on tuvastatav hetkeseisuga 20–25% autismiga patsientidest. Testimise näidustusteks on kliinilise diagnoosi kinnitamine.

Geenide nimekiri (75):

Geen	Fenotüüp
ADNP	Helsmoortel-van der Aa syndrome (Mental retardation, autosomal dominant 28)
BCL11A	Dias-Logan syndrome
C12ORF4	Developmental delay and seizures with or without movement abnormalities (DEDSM)
CACNA1C	Brugada syndrome, Timothy syndrome
CC2D1A	Mental retardation, autosomal recessive 3
CNOT3	Developmental delay and seizures with or without movement abnormalities (DEDSM)
CNTN6	Developmental delay and seizures with or without movement abnormalities (DEDSM)
COL4A3BP	Mental retardation, autosomal dominant 34
CSNK2A1	Jeune asphyxiating thoracic dystrophy, Joubert syndrome 21
CTNND2	Developmental delay and seizures with or without movement abnormalities (DEDSM)
DHCR7	Smith-Lemli-Opitz syndrome
EHMT1	Kleefstra syndrome
EN2	Autism
FBXO11	Developmental delay and seizures with or without movement abnormalities (DEDSM)
FOXP1	Mental retardation with language impairment and autistic features, Congenital heart malformations
GAMT	Guanidinoacetate methyltransferase deficiency
KMT2E	
KMT5B	Developmental delay and seizures with or without movement abnormalities (DEDSM), Autism spectrum disorder, overgrowth syndrome with intellectual disability
MBOAT7	Mental retardation, autosomal recessive 57
MECP2	Angelman-like syndrome, Autism, Rett syndrome, Encephalopathy, Mental retardation

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
NBEA	Epilepsy
NFIB	Macrocephaly
NLGN3	Autism, Asperger syndrome
NLGN4X	Autism, Asperger syndrome, Mental retardation
NSD1	Sotos syndrome, Weaver syndrome, Beckwith-Wiedemann syndrome
POGZ	Mental retardation, autosomal dominant 37 (White-Sutton syndrome)
PTCHD1	Autism susceptibility, X-linked 4
PTEN	Bannayan-Riley-Ruvalcaba syndrome, Lhermitte-Duclos syndrome, Cowden syndrome
RPL10	Autism
SHANK3	Phelan-McDermid syndrome, Schizophrenia 15
TBR1	
TCF20	Developmental delay and seizures with or without movement abnormalities (DEDSM)
TRIP12	Intellectual disability
TSC1	Lymphangioleiomyomatosis, Tuberous sclerosis
TSC2	Lymphangioleiomyomatosis, Tuberous sclerosis

VAMP2	
WASF1	Intellectual disability and seizures
ZSWIM6	Acromelic frontonasal dysostosis

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