
GENETIC TEST:
Ataxia Spasticity (gene panel)

FULL NAME:	Ataxia Spasticity (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region Whole Exome Sequencing (WES) Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	26 Jul 2019 - 16:13
CHANGED:	16 Dec 2022 - 10:27

RELATED CONTENT

Related Laboratories

- [Centrum Medische Genetica - UZ Gent](#)

Related Analytes

- [aladin WD repeat nucleoporin](#)
- [alanyl-tRNA synthetase 1](#)
- [ATP binding cassette subfamily A member 2](#)
- [ATP binding cassette subfamily B member 7](#)
- [ATP binding cassette subfamily D member 1](#)
- [abhydrolase domain containing 12, lysophospholipase](#)
- [ACD shelterin complex subunit and telomerase recruitment factor](#)
- [aconitase 2](#)
- [adenosine deaminase RNA specific](#)
- [adhesion G protein-coupled receptor G1](#)
- [ADP-ribosylserine hydrolase](#)
- [AFG3 like matrix AAA peptidase subunit 2](#)
- [ATP/GTP binding carboxypeptidase 1](#)
- [Abelson helper integration site 1](#)
- [apoptosis inducing factor mitochondria associated 1](#)
- [aminoacyl tRNA synthetase complex interacting multifunctional protein 1](#)
- [aldehyde dehydrogenase 18 family member A1](#)
- [aldehyde dehydrogenase 5 family member A1](#)
- [ALG6 alpha-1,3-glucosyltransferase](#)
- [alsin Rho guanine nucleotide exchange factor ALS2](#)

- adenosine monophosphate deaminase 2
- anoctamin 10
- adaptor related protein complex 4 subunit beta 1
- adaptor related protein complex 4 subunit epsilon 1
- adaptor related protein complex 4 subunit mu 1
- adaptor related protein complex 4 subunit sigma 1
- adaptor related protein complex 5 subunit zeta 1
- apolipoprotein B
- aprataxin
- arginase 1
- ADP ribosylation factor like GTPase 13B
- ADP ribosylation factor like GTPase 6 interacting protein 1
- arylsulfatase A
- arylsulfatase family member I
- aristaless related homeobox
- aspartoacylase
- ATCAY kinesin light chain interacting caytaxin
- autophagy related 5
- atlastin GTPase 1
- ATM serine/threonine kinase
- ATPase cation transporting 13A2
- ATPase Na⁺/K⁺ transporting subunit alpha 3
- ATPase plasma membrane Ca²⁺ transporting 3
- ATPase copper transporting beta
- ATPase phospholipid transporting 8A2
- AU RNA binding methylglutaconyl-CoA hydratase
- beta-1,4-N-acetyl-galactosaminyltransferase 1
- branched chain keto acid dehydrogenase E1 subunit alpha
- branched chain keto acid dehydrogenase E1 subunit beta
- BAF chromatin remodeling complex subunit BCL11B
- BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone
- BICD cargo adaptor 2
- BSCL2 lipid droplet biogenesis associated, seipin

- biotinidase
- chromosome 19 open reading frame 12
- carbonic anhydrase 8
- calcium voltage-gated channel subunit alpha1 A
- calcium voltage-gated channel subunit alpha1 E
- calcium voltage-gated channel subunit alpha1 G
- calcium voltage-gated channel auxiliary subunit beta 4
- calmodulin binding transcription activator 1
- calpain 1
- coiled-coil and C2 domain containing 2A
- coiled-coil domain containing 88C
- chaperonin containing TCP1 subunit 5
- cyclin dependent kinase 16
- centrosomal protein 290
- calcineurin like EF-hand protein 1
- chloride voltage-gated channel 2
- contactin associated protein 1
- cytochrome c oxidase assembly factor 7
- coenzyme Q2, polyprenyltransferase
- coenzyme Q4
- coenzyme Q8A
- coenzyme Q9
- cytochrome c oxidase assembly factor heme A:farnesyltransferase COX10
- cytochrome c oxidase assembly homolog COX15
- cytochrome c oxidase assembly factor COX20
- ceruloplasmin
- carnitine palmitoyltransferase 1C
- cystatin B
- C-terminal binding protein 1
- CWF19 like cell cycle control factor 1
- cytochrome P450 family 27 subfamily A member 1
- cytochrome P450 family 2 subfamily U member 1
- cytochrome P450 family 7 subfamily B member 1

- DAB adaptor protein 1
- aspartyl-tRNA synthetase 1
- aspartyl-tRNA synthetase 2, mitochondrial
- dihydrolipoamide branched chain transacylase E2
- DDHD domain containing 1
- DDHD domain containing 2
- DEAD-box helicase 3 X-linked
- delta 4-desaturase, sphingolipid 1
- dihydrolipoamide S-acetyltransferase
- dihydrolipoamide dehydrogenase
- DnaJ heat shock protein family (Hsp40) member C19
- DnaJ heat shock protein family (Hsp40) member C3
- DnaJ heat shock protein family (Hsp40) member C5
- DNA methyltransferase 1
- dedicator of cytokinesis 3
- glutamyl-tRNA synthetase 2, mitochondrial
- EBF transcription factor 3
- enoyl-CoA hydratase, short chain 1
- eukaryotic translation elongation factor 2
- eukaryotic translation initiation factor 2 alpha kinase 1
- ELOVL fatty acid elongase 1
- ELOVL fatty acid elongase 4
- ELOVL fatty acid elongase 5
- ER membrane protein complex subunit 1
- ectonucleoside triphosphate diphosphohydrolase 1
- EPM2A glucan phosphatase, laforin
- ERCC excision repair 1, endonuclease non-catalytic subunit
- ERCC excision repair 2, TFIIH core complex helicase subunit
- ERCC excision repair 4, endonuclease catalytic subunit
- ERCC excision repair 8, CSA ubiquitin ligase complex subunit
- ER lipid raft associated 1
- ER lipid raft associated 2
- exosome component 3

- fatty acid 2-hydroxylase
- phenylalanyl-tRNA synthetase 2, mitochondrial
- FAST kinase domains 2
- FAT atypical cadherin 2
- fibroblast growth factor 14
- fat storage inducing transmembrane protein 2
- fibronectin leucine rich transmembrane protein 1
- FLVCR heme transporter 1
- folate receptor 1
- FERM domain containing 4A
- FERM domain containing 7
- galactosylceramidase
- gigaxonin
- glucosylceramidase beta 2
- 1,4-alpha-glucan branching enzyme 1
- ganglioside induced differentiation associated protein 2
- glial fibrillary acidic protein
- GTP dependent ribosome recycling factor mitochondrial 2
- gap junction protein gamma 2
- galactosidase beta 1
- glutaredoxin 5
- glutaminase
- ganglioside GM2 activator
- golgi SNAP receptor complex member 2
- G protein-coupled receptor 143
- glutamate ionotropic receptor delta type subunit 2
- glutamate ionotropic receptor NMDA type subunit 1
- glutamate metabotropic receptor 1
- granulin precursor
- GS homeobox 2
- HECT domain and ankyrin repeat containing E3 ubiquitin protein ligase 1
- hexosaminidase subunit alpha
- hexosaminidase subunit beta

- hydroxysteroid 17-beta dehydrogenase 10
- hydroxysteroid 17-beta dehydrogenase 4
- heat shock protein family D (Hsp60) member 1
- iron-sulfur cluster assembly factor IBA57
- interferon induced with helicase C domain 1
- interferon related developmental regulator 1
- intraflagellar transport 140
- integrator complex subunit 8
- interferon regulatory factor 2 binding protein like
- iron-sulfur cluster assembly 1
- integral membrane protein 2B
- inositol 1,4,5-trisphosphate receptor type 1
- junctional adhesion molecule 2
- KN motif and ankyrin repeat domains 1
- potassium voltage-gated channel subfamily A member 1
- potassium voltage-gated channel subfamily A member 2
- potassium voltage-gated channel subfamily A member 4
- potassium voltage-gated channel subfamily C member 3
- potassium voltage-gated channel subfamily D member 3
- potassium inwardly rectifying channel subfamily J member 10
- potassium inwardly rectifying channel subfamily J member 6
- potassium calcium-activated channel subfamily M alpha 1
- potassium voltage-gated channel subfamily Q member 2
- potassium channel tetramerization domain containing 7
- kinase D interacting substrate 220
- kinesin family member 1A
- kinesin family member 1C
- kinesin family member 5A
- kinesin light chain 2
- L1 cell adhesion molecule
- L-2-hydroxyglutarate dehydrogenase
- laminin subunit alpha 1
- laminin subunit beta 1

- lipoyl(octanoyl) transferase 2
- LYR motif containing 7
- lysosomal trafficking regulator
- mannosidase alpha class 2B member 1
- methionyl-tRNA synthetase 1
- methionyl-tRNA synthetase 2, mitochondrial
- mucolipin TRP cation channel 1
- methyl-CpG binding protein 2
- mediator complex subunit 12
- modulator of VRAC current 1
- metabolism of cobalamin associated D
- membrane metalloendopeptidase
- mitochondrial inner membrane protein MPV17
- MRE11 homolog, double strand break repair nuclease
- mitochondrial ribosomal protein S34
- misato mitochondrial distribution and morphology regulator 1
- microtubule crosslinking factor 1
- mitochondrial poly(A) polymerase
- mitochondrial translation release factor in rescue
- microsomal triglyceride transfer protein
- myogenesis regulating glycosidase (putative)
- N-acetylneuraminate synthase
- NADH:ubiquinone oxidoreductase subunit A9
- NADH:ubiquinone oxidoreductase complex assembly factor 4
- NADH:ubiquinone oxidoreductase complex assembly factor 6
- NADH:ubiquinone oxidoreductase core subunit S1
- NADH:ubiquinone oxidoreductase core subunit S3
- NADH:ubiquinone oxidoreductase core subunit S7
- neurofilament light chain
- neuraminidase 1
- neurite extension and migration factor
- NF2, moesin-ezrin-radixin like (MERLIN) tumor suppressor
- NHL repeat containing E3 ubiquitin protein ligase 1

- NIPA magnesium transporter 1
- NK6 homeobox 2
- nucleolar protein 3
- NPC intracellular cholesterol transporter 1
- NPC intracellular cholesterol transporter 2
- 5'-nucleotidase, cytosolic II
- netrin G2
- nucleoporin 93
- NUS1 dehydrodolichyl diphosphate synthase subunit
- occludin
- OFD1 centriole and centriolar satellite protein
- outer mitochondrial membrane lipid metabolism regulator OPA3
- oligophrenin 1
- ornithine transcarbamylase
- prolyl-tRNA synthetase 2, mitochondrial
- protocadherin 12
- phosphate cytidylyltransferase 2, ethanolamine
- pyruvate dehydrogenase E1 subunit alpha 1
- pyruvate dehydrogenase complex component X
- prodynorphin
- peroxisomal biogenesis factor 10
- peroxisomal biogenesis factor 16
- peroxisomal biogenesis factor 7
- post-GPI attachment to proteins inositol deacylase 1
- phytanoyl-CoA 2-hydroxylase
- phosphoinositide-3-kinase regulatory subunit 5
- phospholipase A2 group VI
- phospholipase A2 activating protein
- phospholipase D family member 3
- proteolipid protein 1
- phosphomannomutase 2
- peptidase, mitochondrial processing subunit alpha
- peptidase, mitochondrial processing subunit beta

- polynucleotide kinase 3'-phosphatase
- patatin like phospholipase domain containing 6
- podocalyxin like
- DNA polymerase gamma, catalytic subunit
- RNA polymerase III subunit A
- RNA polymerase III subunit B
- perforin 1
- protein kinase C gamma
- prion protein
- phosphoribosyl pyrophosphate synthetase 1
- proline rich transmembrane protein 2
- prosaposin
- presenilin 1
- protein tyrosine phosphatase non-receptor type 23
- peptidyl-tRNA hydrolase 2
- pumilio RNA binding family member 1
- RAB11B, member RAS oncogene family
- RAB18, member RAS oncogene family
- RAB3 GTPase activating protein catalytic subunit 1
- RAB3 GTPase activating non-catalytic protein subunit 2
- receptor accessory protein 1
- receptor accessory protein 2
- reticulophagy regulator 1
- ribonuclease H2 subunit A
- ribonuclease H2 subunit B
- ribonuclease H2 subunit C
- ring finger protein 168
- ring finger protein 170
- ring finger protein 216
- RAR related orphan receptor A
- RPGRIP1 like
- reticulon 2
- reticulon 4 interacting protein 1

- rubicon autophagy regulator
- sacsin molecular chaperone
- sterile alpha motif domain containing 9 like
- SAM and HD domain containing deoxynucleoside triphosphate triphosphohydrolase 1
- seryl-tRNA synthetase 1
- sodium voltage-gated channel alpha subunit 8
- SCY1 like pseudokinase 1
- succinate dehydrogenase complex flavoprotein subunit A
- succinate dehydrogenase complex assembly factor 1
- succinate dehydrogenase complex subunit D
- Sep (O-phosphoserine) tRNA:Sec (selenocysteine) tRNA synthase
- senataxin
- SIL1 nucleotide exchange factor
- solute carrier family 13 member 5
- solute carrier family 16 member 2
- solute carrier family 17 member 5
- solute carrier family 19 member 3
- solute carrier family 1 member 3
- solute carrier family 1 member 4
- solute carrier family 25 member 15
- solute carrier family 25 member 46
- solute carrier family 2 member 1
- solute carrier family 33 member 1
- solute carrier family 44 member 1
- solute carrier family 52 member 2
- solute carrier family 52 member 3
- solute carrier family 6 member 8
- solute carrier family 9 member A1
- solute carrier family 9 member A6
- sphingomyelin phosphodiesterase 1
- synaptosome associated protein 25
- sorting nexin 14
- SRY-box transcription factor 10

- spartin
- spastin
- SPG11 vesicle trafficking associated, spatacsin
- SPG21 abhydrolase domain containing, maspardin
- SPG7 matrix AAA peptidase subunit, paraplegin
- sepiapterin reductase
- spectrin alpha, non-erythrocytic 1
- spectrin beta, non-erythrocytic 2
- sequestosome 1
- steroid 5 alpha-reductase 3
- STIP1 homology and U-box containing protein 1
- succinate-CoA ligase ADP-forming subunit beta
- sulfite oxidase
- SURF1 cytochrome c oxidase assembly factor
- small vasohibin binding protein
- spectrin repeat containing nuclear envelope protein 1
- synaptotagmin 14
- transport and golgi organization 2 homolog
- TBC1 domain family member 20
- tyrosyl-DNA phosphodiesterase 1
- tyrosyl-DNA phosphodiesterase 2
- tectonin beta-propeller repeat containing 2
- teneurin transmembrane protein 3
- trafficking from ER to golgi regulator
- transglutaminase 6
- tRNA-histidine guanylyltransferase 1 like
- transmembrane protein 216
- transmembrane protein 231
- transmembrane protein 240
- transmembrane protein 67
- thioredoxin related transmembrane protein 2
- trafficking kinesin protein 1
- trafficking protein particle complex subunit 12

- trafficking protein particle complex subunit 4
- tRNA isopentenyltransferase 1
- transient receptor potential cation channel subfamily C member 3
- tRNA splicing endonuclease subunit 2
- tRNA splicing endonuclease subunit 54
- tau tubulin kinase 2
- tetratricopeptide repeat domain 19
- alpha tocopherol transfer protein
- tubulin alpha 1a
- tubulin beta 4A class IVa
- tubulin gamma 1
- twinkle mtDNA helicase
- ubiquitin like modifier activating enzyme 5
- ubiquitin associated protein 1
- upstream binding transcription factor
- ubiquitin C-terminal hydrolase L1
- ubiquitin-fold modifier conjugating enzyme 1
- ubiquitin fold modifier 1
- UDP-glucose pyrophosphorylase 2
- ubiquitin specific peptidase 8
- vesicle associated membrane protein 1
- valosin containing protein
- very low density lipoprotein receptor
- vacuolar protein sorting 13 homolog B
- vacuolar protein sorting 13 homolog D
- VPS37A subunit of ESCRT-I
- VRK serine/threonine kinase 1
- von Willebrand factor A domain containing 3B
- tryptophanyl tRNA synthetase 2, mitochondrial
- WASH complex subunit 5
- WD repeat domain 45B
- WD repeat domain 48
- WD repeat domain 73

- WD repeat domain 81
- WW domain containing oxidoreductase
- X-ray repair cross complementing 1
- zinc finger C4H2-type containing
- zinc finger RNA binding protein
- zinc finger FYVE-type containing 26
- zinc finger FYVE-type containing 27

Related Gene Panels

- Ataxia Spasticity - UGent

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