

GENETIC TEST:**Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)**

FULL NAME:	Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)
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)
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-03-25 / 2024-09-09
EQA:	• Ataxia and spastic paraplegias
TURNAROUND TIME (MAXIMUM):	6 months

CREATED:	22 Aug 2019 - 09:23
CHANGED:	19 Jan 2024 - 09:06
URL:	https://ulbgenetics.be/compendium-des-analyses/#GM08

Source URL: http://gentest.healthdata.be/genetic_test/581

RELATED CONTENT

Related Laboratories

- [Centre de Génétique Humaine - Erasme ULB](#)

Related Analytes

- [aladin WD repeat nucleoporin](#)
- [ATP binding cassette subfamily B member 7](#)
- [ATP binding cassette subfamily D member 1](#)
- [abhydrolase domain containing 12, lysophospholipase](#)
- [abhydrolase domain containing 5, lysophosphatidic acid acyltransferase](#)
- [acyl-CoA dehydrogenase very long chain](#)
- [ACD shelterin complex subunit and telomerase recruitment factor](#)
- [aconitase 2](#)
- [adhesion G protein-coupled receptor G1](#)
- [AFG3 like matrix AAA peptidase subunit 2](#)
- [Abelson helper integration site 1](#)
- [5'-aminolevulinate synthase 2](#)
- [aldehyde dehydrogenase 18 family member A1](#)
- [aldehyde dehydrogenase 5 family member A1](#)
- [ALG6 alpha-1,3-glucosyltransferase](#)
- [alsin Rho guanine nucleotide exchange factor ALS2](#)
- [alpha-methylacyl-CoA racemase](#)
- [adenosine monophosphate deaminase 2](#)
- [aminomethyltransferase](#)
- [anoctamin 10](#)

- adaptor related protein complex 1 subunit sigma 2
- aprataxin
- ADP ribosylation factor like GTPase 13B
- ADP ribosylation factor like GTPase 6
- arylsulfatase A
- aristaless related homeobox
- aspartoacylase
- ATCAY kinesin light chain interacting caytaxin
- autophagy related 5
- atlastin GTPase 1
- ATM serine/threonine kinase
- ATPase Na⁺/K⁺ transporting subunit alpha 3
- ATPase plasma membrane Ca²⁺ transporting 2
- ATPase plasma membrane Ca²⁺ transporting 3
- ATPase copper transporting beta
- ATPase phospholipid transporting 8A2
- ataxin 1
- ataxin 10
- ataxin 2
- ataxin 3
- ataxin 7
- B9 domain containing 1
- Bardet-Biedl syndrome 1
- Bardet-Biedl syndrome 10
- Bardet-Biedl syndrome 12
- Bardet-Biedl syndrome 2
- Bardet-Biedl syndrome 4
- Bardet-Biedl syndrome 5
- Bardet-Biedl syndrome 7
- Bardet-Biedl syndrome 9
- brain expressed associated with NEDD4 1
- basic, immunoglobulin-like variable motif containing
- 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 G
- calcium voltage-gated channel auxiliary subunit beta 4
- calmodulin binding transcription activator 1
- calpain 1
- calcium/calmodulin dependent serine protein kinase
- coiled-coil and C2 domain containing 2A
- coiled-coil domain containing 88C
- centrosomal protein 290
- centrosomal protein 41
- charged multivesicular body protein 1A
- chloride voltage-gated channel 2
- CLN5 intracellular trafficking protein
- CLN6 transmembrane ER protein
- caseinolytic mitochondrial matrix peptidase proteolytic subunit
- cytochrome c oxidase assembly factor 7
- Coenzyme A synthase
- component of oligomeric golgi complex 5
- coenzyme Q2, polyprenyltransferase
- coenzyme Q6, monooxygenase
- coenzyme Q8A
- coenzyme Q9
- cytochrome c oxidase assembly factor COX20
- ceruloplasmin
- ciliogenesis and planar polarity effector complex subunit 1
- carnitine acetyltransferase
- cystatin B
- 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
- aspartyl-tRNA synthetase 1
- aspartyl-tRNA synthetase 2, mitochondrial
- damage specific DNA binding protein 2
- DDHD domain containing 2
- dihydrolipoamide S-acetyltransferase
- Dmx like 2
- 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
- EBF transcription factor 3
- eukaryotic translation elongation factor 2
- eukaryotic translation initiation factor 2B subunit alpha
- eukaryotic translation initiation factor 2B subunit beta
- eukaryotic translation initiation factor 2B subunit gamma
- eukaryotic translation initiation factor 2B subunit delta
- eukaryotic translation initiation factor 2B subunit epsilon
- ELOVL fatty acid elongase 4
- ELOVL fatty acid elongase 5
- EPM2A glucan phosphatase, laforin
- ERCC excision repair 2, TFIIH core complex helicase subunit
- ERCC excision repair 3, TFIIH core complex helicase subunit
- ERCC excision repair 4, endonuclease catalytic subunit
- ERCC excision repair 5, endonuclease
- electron transfer flavoprotein subunit alpha
- electron transfer flavoprotein subunit beta
- electron transfer flavoprotein dehydrogenase
- exosome component 3
- fatty acid 2-hydroxylase
- phenylalanyl-tRNA synthetase 2, mitochondrial
- F-box and leucine rich repeat protein 4
- ferredoxin reductase

- fibroblast growth factor 14
- FLVCR heme transporter 1
- fragile X messenger ribonucleoprotein 1
- folate receptor 1
- FERM domain containing 7
- frataxin
- galactosylceramidase
- glucosylceramidase beta 1
- glucosylceramidase beta 2
- 1,4-alpha-glucan branching enzyme 1
- glutaryl-CoA dehydrogenase
- glutamate-cysteine ligase catalytic subunit
- glycine cleavage system protein H
- glial fibrillary acidic protein
- gap junction protein gamma 2
- galactosidase beta 1
- glycine decarboxylase
- glutaredoxin 5
- ganglioside GM2 activator
- golgi SNAP receptor complex member 2
- glycosylphosphatidylinositol anchor attachment 1
- G protein-coupled receptor 143
- glutamate ionotropic receptor delta type subunit 2
- glutamate metabotropic receptor 1
- glutathione synthetase
- histidyl-tRNA synthetase 2, mitochondrial
- hepatic and glial cell adhesion molecule
- HECT and RLD domain containing E3 ubiquitin protein ligase family member 1
- hexosaminidase subunit alpha
- hexosaminidase subunit beta
- 3-hydroxyisobutyryl-CoA hydrolase
- hypoxanthine phosphoribosyltransferase 1
- HtrA serine peptidase 1

- interferon related developmental regulator 1
- immunoglobulin superfamily DCC subclass member 3
- inositol polyphosphate-5-phosphatase E
- interferon regulatory factor 2 binding protein like
- integral membrane protein 2B
- inositol 1,4,5-trisphosphate receptor type 1
- potassium voltage-gated channel subfamily A member 1
- 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
- kinesin family member 1A
- kinesin family member 1C
- kinesin family member 5A
- kinesin family member 5C
- kinesin family member 7
- L-2-hydroxyglutarate dehydrogenase
- laminin subunit alpha 1
- laminin subunit beta 1
- leucyl-tRNA synthetase 2, mitochondrial
- lamin B1
- leucine rich pentatricopeptide repeat containing
- lysosomal trafficking regulator
- mediator complex subunit 12
- major facilitator superfamily domain containing 8
- MKKS centrosomal shuttling protein
- MKS transition zone complex subunit 1
- modulator of VRAC current 1
- metabolism of cobalamin associated C
- membrane metalloendopeptidase
- MRE11 homolog, double strand break repair nuclease
- misato mitochondrial distribution and morphology regulator 1

- mitochondrial methionyl-tRNA formyltransferase
- mitochondrial poly(A) polymerase
- mitochondrial translation release factor in rescue
- 5-methyltetrahydrofolate-homocysteine methyltransferase reductase
- microsomal triglyceride transfer protein
- mevalonate kinase
- N-acetylneuraminate synthase
- NADH:ubiquinone oxidoreductase complex assembly factor 6
- NADH:ubiquinone oxidoreductase core subunit S1
- NADH:ubiquinone oxidoreductase core subunit S2
- NADH:ubiquinone oxidoreductase subunit S4
- NADH:ubiquinone oxidoreductase core subunit S7
- NADH:ubiquinone oxidoreductase core subunit S8
- NADH:ubiquinone oxidoreductase core subunit V1
- neurofilament light chain
- neuraminidase 1
- neurite extension and migration factor
- NHL repeat containing E3 ubiquitin protein ligase 1
- NK6 homeobox 2
- nucleolar protein 3
- NOP56 ribonucleoprotein
- notch receptor 3
- NPC intracellular cholesterol transporter 1
- NPC intracellular cholesterol transporter 2
- nephrocystin 1
- NUBP iron-sulfur cluster assembly factor, mitochondrial
- occludin
- OFD1 centriole and centriolar satellite protein
- outer mitochondrial membrane lipid metabolism regulator OPA3
- oligophrenin 1
- OTU deubiquitinase 4
- paired box 6
- platelet derived growth factor receptor beta

- pyruvate dehydrogenase complex component X
- decaprenyl diphosphate synthase subunit 1
- decaprenyl diphosphate synthase subunit 2
- prodynorphin
- peroxisomal biogenesis factor 10
- peroxisomal biogenesis factor 16
- peroxisomal biogenesis factor 2
- peroxisomal biogenesis factor 7
- phytanoyl-CoA 2-hydroxylase
- phosphoinositide-3-kinase regulatory subunit 5
- phospholipase A2 group VI
- pleckstrin homology and RhoGEF domain containing G4
- proteolipid protein 1
- phosphomannomutase 2
- peptidase, mitochondrial processing subunit alpha
- PNKD metallo-beta-lactamase domain containing
- polynucleotide kinase 3'-phosphatase
- patatin like phospholipase domain containing 6
- DNA polymerase gamma, catalytic subunit
- DNA polymerase eta
- RNA polymerase III subunit A
- RNA polymerase III subunit B
- protein phosphatase 2 regulatory subunit Bbeta
- prickle planar cell polarity protein 1
- protein kinase C gamma
- prion protein
- phosphoribosyl pyrophosphate synthetase 1
- proline rich transmembrane protein 2
- phosphatase and tensin homolog
- pumilio RNA binding family member 1
- RAB18, member RAS oncogene family
- RAB3A, member RAS oncogene family
- RAB3 GTPase activating protein catalytic subunit 1

- arginyl-tRNA synthetase 2, mitochondrial
- reelin
- ribonuclease H2 subunit A
- ribonuclease H2 subunit C
- ring finger protein 170
- ring finger protein 216
- RPGRIP1 like
- ribonucleotide reductase regulatory TP53 inducible subunit M2B
- rubicon autophagy regulator
- sacsin molecular chaperone
- sterile alpha motif domain containing 9 like
- SAM and HD domain containing deoxynucleoside triphosphate triphosphohydrolase 1
- secretion associated Ras related GTPase 1B
- sodium voltage-gated channel alpha subunit 8
- SEC16 homolog A, endoplasmic reticulum export factor
- Sep (O-phosphoserine) tRNA:Sec (selenocysteine) tRNA synthase
- serine active site containing 1
- senataxin
- SIL1 nucleotide exchange factor
- SKI family transcriptional corepressor 1
- 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 20 member 2
- solute carrier family 25 member 15
- solute carrier family 25 member 46
- solute carrier family 2 member 1
- solute carrier family 39 member 8
- solute carrier family 52 member 2
- solute carrier family 52 member 3
- solute carrier family 9 member A1
- solute carrier family 9 member A6

- sphingomyelin phosphodiesterase 1
- sorting nexin 14
- spastin
- SPG11 vesicle trafficking associated, spatacsin
- SPG7 matrix AAA peptidase subunit, paraplegin
- sepiapterin reductase
- spectrin beta, non-erythrocytic 2
- steroid 5 alpha-reductase 3
- STIP1 homology and U-box containing protein 1
- spectrin repeat containing nuclear envelope protein 1
- synaptotagmin 14
- transport and golgi organization 2 homolog
- TBC1 domain family member 20
- tubulin folding cofactor C
- TATA-box binding protein
- tectonic family member 1
- tectonic family member 2
- tectonic family member 3
- tyrosyl-DNA phosphodiesterase 1
- tyrosyl-DNA phosphodiesterase 2
- transglutaminase 6
- thymidine kinase 2
- transmembrane protein 138
- transmembrane protein 216
- transmembrane protein 231
- transmembrane protein 237
- transmembrane protein 240
- transmembrane protein 67
- tripartite motif containing 32
- transient receptor potential cation channel subfamily C member 3
- tRNA splicing endonuclease subunit 2
- tRNA splicing endonuclease subunit 34
- tRNA splicing endonuclease subunit 54

- Ts translation elongation factor, mitochondrial
- tau tubulin kinase 2
- tetratricopeptide repeat domain 19
- tetratricopeptide repeat domain 8
- alpha tocopherol transfer protein
- tubulin beta 4A class IVa
- tubulin gamma 1
- twinkle mtDNA helicase
- thymidine phosphorylase
- ubiquitin like modifier activating enzyme 5
- vesicle associated membrane protein 1
- very low density lipoprotein receptor
- vacuolar protein sorting 13 homolog A
- vacuolar protein sorting 13 homolog B
- vacuolar protein sorting 13 homolog D
- VRK serine/threonine kinase 1
- von Willebrand factor A domain containing 3B
- WASH complex subunit 5
- WD repeat containing planar cell polarity effector
- WD repeat domain 73
- WD repeat domain 81
- wolframin ER transmembrane glycoprotein
- WW domain containing oxidoreductase
- XPA, DNA damage recognition and repair factor
- XPC complex subunit, DNA damage recognition and repair factor
- xenotropic and polytropic retrovirus receptor 1
- X-ray repair cross complementing 1
- zinc finger C4H2-type containing
- zinc finger FYVE-type containing 26
- zinc finger protein 423
- zinc finger protein 592

Related Gene Panels

- [Ataxia \(348 genes\) - ULB](#)

Source URL: http://gentest.healthdata.be/genetic_test/581