GENETIC TEST:
Hereditary Spastic Paraplegia (gene panel)

FULL NAME:	Hereditary Spastic Paraplegia (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, DNA
METHOD CATEGORY:	Sequence analysis: entire coding region Whole Exome Sequencing (WES)
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2023-11-09 / 2024-05-08
EQA:	• Ataxia and spastic paraplegias
TURNAROUND TIME (MAXIMUM):	6 months

CREATED:	11 Dec 2020 - 09:38
CHANGED:	22 Jan 2024 - 13:50
URL:	https://labogidsmedgen.uza.be/analyses/hereditaire-spastische-paraplegie-hsp

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RELATED CONTENT

Related Diseases

- Ataxia with vitamin E deficiency
- Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome
- Autosomal dominant spastic paraplegia type 10
- Autosomal dominant spastic paraplegia type 12
- Autosomal dominant spastic paraplegia type 17
- Autosomal dominant spastic paraplegia type 3
- Autosomal dominant spastic paraplegia type 31
- Autosomal dominant spastic paraplegia type 4
- Autosomal dominant spastic paraplegia type 73
- Autosomal dominant spastic paraplegia type 8
- Autosomal dominant spastic paraplegia type 9A
- Autosomal dominant spastic paraplegia type 9B
- Autosomal recessive ataxia, Beauce type
- Autosomal recessive cerebellar ataxia due to STUB1 deficiency
- Autosomal recessive cerebellar ataxia with late-onset spasticity
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay
- Autosomal recessive spastic paraplegia type 11
- Autosomal recessive spastic paraplegia type 15
- Autosomal recessive spastic paraplegia type 21
- Autosomal recessive spastic paraplegia type 26
- Autosomal recessive spastic paraplegia type 28
- Autosomal recessive spastic paraplegia type 35
- Autosomal recessive spastic paraplegia type 39
- Autosomal recessive spastic paraplegia type 46
- Autosomal recessive spastic paraplegia type 48
- Autosomal recessive spastic paraplegia type 5A

- Autosomal recessive spastic paraplegia type 62
- Autosomal recessive spastic paraplegia type 76
- Autosomal recessive spastic paraplegia type 77
- Autosomal recessive spastic paraplegia type 78
- Autosomal recessive spastic paraplegia type 9B
- Autosomal spastic paraplegia type 18
- Autosomal spastic paraplegia type 30
- Autosomal spastic paraplegia type 58
- Autosomal spastic paraplegia type 72
- Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome
- Severe intellectual disability and progressive spastic paraplegia
- Spastic paraplegia type 2
- Spastic paraplegia-severe developmental delay-epilepsy syndrome
- Spectrin-associated autosomal recessive cerebellar ataxia
- Spinocerebellar ataxia with axonal neuropathy type 2
- Wilson disease
- X-linked complicated spastic paraplegia type 1

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

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 16A, phospholipase
- AFG3 like matrix AAA peptidase subunit 2

- apoptosis inducing factor mitochondria associated 1
- aminoacyl tRNA synthetase complex interacting multifunctional protein 1
- aldehyde dehydrogenase 18 family member A1
- alsin Rho guanine nucleotide exchange factor ALS2
- adenosine monophosphate deaminase 2
- anoctamin 10
- adaptor related protein complex 1 subunit sigma 2
- 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
- aprataxin
- arginase 1
- ADP ribosylation factor like GTPase 6 interacting protein 1
- arylsulfatase A
- ATCAY kinesin light chain interacting caytaxin
- autophagy related 7
- atlastin GTPase 1
- ATM serine/threonine kinase
- ATPase cation transporting 13A2
- ATPase Na+/K+ transporting subunit alpha 3
- ATPase plasma membrane Ca2+ transporting 3
- ATP synthase membrane subunit c locus 3
- ATPase copper transporting beta
- ATPase phospholipid transporting 8A2
- AU RNA binding methylglutaconyl-CoA hydratase
- beta-1,4-N-acetyl-galactosaminyltransferase 1
- 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 alpha2delta 2
- calmodulin binding transcription activator 1
- calpain 1
- calcium/calmodulin dependent serine protein kinase
- chaperonin containing TCP1 subunit 5
- charged multivesicular body protein 1A
- chloride voltage-gated channel 2
- CLN6 transmembrane ER protein
- cleavage factor polyribonucleotide kinase subunit 1
- cytochrome c oxidase assembly factor 7
- Coenzyme A synthase
- component of oligomeric golgi complex 5
- coenzyme Q8A
- cytochrome c oxidase assembly factor COX20
- carnitine palmitoyltransferase 1C
- catenin beta 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
- aspartyl-tRNA synthetase 1
- aspartyl-tRNA synthetase 2, mitochondrial
- DDHD domain containing 1
- DDHD domain containing 2
- DnaJ heat shock protein family (Hsp40) member C5
- DNA methyltransferase 1
- 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
- ectonucleoside triphosphate diphosphohydrolase 1
- ER lipid raft associated 1
- ER lipid raft associated 2
- exosome component 3
- exosome component 8
- exosome component 9
- fatty acid 2-hydroxylase
- fatty acyl-CoA reductase 1
- phenylalanyl-tRNA synthetase 2, mitochondrial
- FAT atypical cadherin 2
- fibroblast growth factor 14
- FLVCR heme transporter 1
- galactosylceramidase
- glucosylceramidase beta 2
- GTP cyclohydrolase 1
- ganglioside induced differentiation associated protein 2
- glial fibrillary acidic protein
- gap junction protein gamma 2
- glutaredoxin 5
- golgi SNAP receptor complex member 2
- glycosylphosphatidylinositol anchor attachment 1
- glutamate ionotropic receptor delta type subunit 2
- glutamate metabotropic receptor 1
- HECT domain and ankyrin repeat containing E3 ubiquitin protein ligase 1
- hexosaminidase subunit alpha
- hexosaminidase subunit beta
- heat shock protein nuclear import factor hikeshi
- 4-hydroxyphenylpyruvate dioxygenase like
- heat shock protein family D (Hsp60) member 1
- iron-sulfur cluster assembly factor IBA57
- interferon related developmental regulator 1

- inositol polyphosphate-5-phosphatase E
- interferon regulatory factor 2 binding protein like
- inositol 1,4,5-trisphosphate receptor type 1
- potassium voltage-gated channel subfamily A member 1
- potassium voltage-gated channel subfamily A member 2
- potassium voltage-gated channel subfamily C member 3
- potassium voltage-gated channel subfamily D member 3
- kinase D interacting substrate 220
- kinesin family member 1A
- kinesin family member 1C
- kinesin family member 5A
- karyopherin subunit alpha 3
- L1 cell adhesion molecule
- myelin associated glycoprotein
- methionyl-tRNA synthetase 2, mitochondrial
- metabolism of cobalamin associated C
- MRE11 homolog, double strand break repair nuclease
- misato mitochondrial distribution and morphology regulator 1
- mitochondrial translation release factor in rescue
- microsomal triglyceride transfer protein
- NIPA magnesium transporter 1
- NK6 homeobox 2
- NPC intracellular cholesterol transporter 1
- NPC intracellular cholesterol transporter 2
- neuronal pentraxin 1
- 5'-nucleotidase, cytosolic II
- OPA1 mitochondrial dynamin like GTPase
- outer mitochondrial membrane lipid metabolism regulator OPA3
- protocadherin 12
- phosphate cytidylyltransferase 2, ethanolamine
- prodynorphin
- peroxisomal biogenesis factor 7
- post-GPI attachment to proteins inositol deacylase 1

- phytanoyl-CoA 2-hydroxylase
- phospholipase A2 group VI
- proteolipid protein 1
- peptidase, mitochondrial processing subunit alpha
- peptidase, mitochondrial processing subunit beta
- polynucleotide kinase 3'-phosphatase
- patatin like phospholipase domain containing 6
- DNA polymerase gamma, catalytic subunit
- RNA polymerase III subunit A
- prickle planar cell polarity protein 1
- protein kinase C gamma
- pumilio RNA binding family member 1
- arginyl-tRNA synthetase 2, mitochondrial
- receptor accessory protein 1
- receptor accessory protein 2
- replication factor C subunit 1
- ring finger protein 170
- ring finger protein 216
- ring finger protein 220
- reticulon 2
- rubicon autophagy regulator
- sacsin molecular chaperone
- sterile alpha motif domain containing 9 like
- sodium voltage-gated channel alpha subunit 8
- SCY1 like pseudokinase 1
- selenoprotein I
- Sep (O-phosphoserine) tRNA:Sec (selenocysteine) tRNA synthase
- senataxin
- SIL1 nucleotide exchange factor
- solute carrier family 1 member 3
- solute carrier family 1 member 4
- solute carrier family 25 member 46
- solute carrier family 33 member 1

- solute carrier family 9 member A1
- sorting nexin 14
- spartin
- spastin
- SPG11 vesicle trafficking associated, spatacsin
- SPG21 abhydrolase domain containing, maspardin
- SPG7 matrix AAA peptidase subunit, paraplegin
- spectrin alpha, non-erythrocytic 1
- spectrin beta, non-erythrocytic 2
- sequestosome 1
- STIP1 homology and U-box containing protein 1
- spectrin repeat containing nuclear envelope protein 1
- tyrosyl-DNA phosphodiesterase 2
- trafficking from ER to golgi regulator
- transglutaminase 6
- tRNA-histidine guanylyltransferase 1 like
- transmembrane protein 240
- transmembrane protein 63C
- target of EGR1, exonuclease
- tripeptidyl peptidase 1
- tRNA splicing endonuclease subunit 2
- tRNA splicing endonuclease subunit 54
- tau tubulin kinase 2
- alpha tocopherol transfer protein
- tubulin beta 4A class IVa
- ubiquitin associated protein 1
- ubiquitin C-terminal hydrolase L1
- vesicle associated membrane protein 1
- very low density lipoprotein receptor
- vacuolar protein sorting 13 homolog D
- VPS41 subunit of HOPS complex
- VPS53 subunit of GARP complex
- VRK serine/threonine kinase 1

- WASH complex subunit 5
- WD repeat domain 45B
- WD repeat domain 73
- WD repeat domain 81
- zinc finger FYVE-type containing 26

Related Gene Panels

- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
