GENETIC TEST:
Hereditary Spastic Paraplegia (94 genes)

FULL NAME:	Hereditary Spastic Paraplegia (94 genes)
DESCRIPTION:	HSP panel (94 genen): ABCD1, ADAR, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, ATL1, ATP13A2, B4GALNT1, BICD2, BSCL2, C12orf65, C19orf12, CAPN1, CCT5, CPT1C, CYP27A1, CYP2U1, CYP7B1, DARS, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FLRT1, GBA2, GCH1, GJC2, HACE1, HSPD1, IBA57, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, L1CAM, MAG, MARS, MARS2, MTPAP, NIPA1, NKX6-2, NT5C2, OPA3, PCYT2, PGAP1, PLP1, PNPLA6, POLR3A, RAB3GAP2, REEP1, REEP2, RNASEH2B, RTN2, SACS, SERAC1, SLC16A2, SLC1A4, SLC2A1, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TUBB4A, UBAP1, UCHL1, USP8, VAMP1, VPS37A, WASHC5, WDR48, ZFR, ZFYVE26, ZFYVE27
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

METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
TURNAROUND TIME (MAXIMUM):	6 - 9 months
CREATED:	19 Jul 2019 - 12:39
CHANGED:	15 Oct 2021 - 14:33
URL:	https://laboboeken.nexuzhealth.com/pboek/internet/GHB%20CME/15334

RELATED CONTENT

Related Diseases

- Hereditary spastic paraplegia

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ATP binding cassette subfamily D member 1
- adenosine deaminase RNA specific
- AFG3 like matrix AAA peptidase subunit 2
- aminoacyl tRNA synthetase complex interacting multifunctional protein 1
- aldehyde dehydrogenase 18 family member A1
- aldehyde dehydrogenase 3 family member A2
- alsin Rho guanine nucleotide exchange factor ALS2
- adenosine monophosphate deaminase 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
- ADP ribosylation factor like GTPase 6 interacting protein 1
- arylsulfatase family member I

- atlastin GTPase 1
- ATPase cation transporting 13A2
- beta-1,4-N-acetyl-galactosaminyltransferase 1
- BICD cargo adaptor 2
- B-Raf proto-oncogene, serine/threonine kinase
- BSCL2 lipid droplet biogenesis associated, seipin
- chromosome 19 open reading frame 12
- calpain 1
- Cbl proto-oncogene
- chaperonin containing TCP1 subunit 5
- carnitine palmitoyltransferase 1C
- 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
- DDHD domain containing 1
- DDHD domain containing 2
- dual serine/threonine and tyrosine protein kinase
- ectonucleoside triphosphate diphosphohydrolase 1
- ERCC excision repair 2, TFIIH core complex helicase subunit
- ER lipid raft associated 1
- ER lipid raft associated 2
- fatty acid 2-hydroxylase
- phenylalanyl-tRNA synthetase 2, mitochondrial
- fibronectin leucine rich transmembrane protein 1
- glucosylceramidase beta 2
- GTP cyclohydrolase 1
- gap junction protein gamma 2
- HECT domain and ankyrin repeat containing E3 ubiquitin protein ligase 1
- HRas proto-oncogene, GTPase
- heat shock protein family D (Hsp60) member 1
- iron-sulfur cluster assembly factor IBA57
- potassium voltage-gated channel subfamily A member 2

- lysine demethylase 5C
- kinase D interacting substrate 220
- kinesin family member 1A
- kinesin family member 1C
- kinesin family member 5A
- kinesin light chain 2
- KRAS proto-oncogene, GTPase
- L1 cell adhesion molecule
- mitogen-activated protein kinase kinase 1
- mitogen-activated protein kinase kinase 2
- methionyl-tRNA synthetase 1
- methionyl-tRNA synthetase 2, mitochondrial
- mitochondrial poly(A) polymerase
- mitochondrial translation release factor in rescue
- NIPA magnesium transporter 1
- NK6 homeobox 2
- NRAS proto-oncogene, GTPase
- 5'-nucleotidase, cytosolic II
- outer mitochondrial membrane lipid metabolism regulator OPA3
- phosphate cytidylyltransferase 2, ethanolamine
- post-GPI attachment to proteins inositol deacylase 1
- proteolipid protein 1
- patatin like phospholipase domain containing 6
- RNA polymerase III subunit A
- protein tyrosine phosphatase non-receptor type 11
- RAB3 GTPase activating non-catalytic protein subunit 2
- Raf-1 proto-oncogene, serine/threonine kinase
- receptor accessory protein 1
- receptor accessory protein 2
- Ras like without CAAX 1
- ribonuclease H2 subunit B
- reticulon 2
- sacsin molecular chaperone

- serine active site containing 1
- SHOC2 leucine rich repeat scaffold protein
- solute carrier family 16 member 2
- solute carrier family 1 member 4
- solute carrier family 2 member 1
- solute carrier family 33 member 1
- SOS Ras/Rac guanine nucleotide exchange factor 1
- spartin
- spastin
- SPG11 vesicle trafficking associated, spatacsin
- SPG21 abhydrolase domain containing, maspardin
- SPG7 matrix AAA peptidase subunit, paraplegin
- tectonin beta-propeller repeat containing 2
- trafficking from ER to golgi regulator
- tubulin beta 4A class IVa
- ubiquitin associated protein 1
- ubiquitin C-terminal hydrolase L1
- ubiquitin specific peptidase 8
- vesicle associated membrane protein 1
- VPS37A subunit of ESCRT-I
- WASH complex subunit 5
- WD repeat domain 48
- zinc finger RNA binding protein
- zinc finger FYVE-type containing 26
- zinc finger FYVE-type containing 27

Related Gene Panels

- Hereditary Spastic Paraplegia (94 genes) - KUL
