GENETIC TEST:
Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)

FULL NAME:	Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
DESCRIPTION:	Charcot-Marie-Tooth (other than type 1A) (139 genes, IPN panel) AARS, ABCD1, ABHD12, AGTPBP1, AIFM1, APTX, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, C12orf65, CCT5, CD59, CNTNAP1, COA7, COX6A1, CTDP1, DCAF8, DCTN1, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, GAN, GARS, GDAP1, GJB1, GJB3, GLA, GNB4, HADHA, HADHB, HARS, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IARS2, IFRD1, IGHMBP2, INF2, ITPR3, KARS, KIF1A, KIF1B, KIF5A, KLHL13, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, OPA3, PDK3, PDXK, PEX1, PEX7, PHYH, PLEKHG5, PMP2, PMP22, PNKP, POLG, PRDM12, PRNP, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SCO2, SCP2, SEPT9, SETX, SGPL1, SH3BP4, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A7, SORD, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, TDP1, TECPR2, TFG, TRIM2, TRPA1, TRPV4, TTR, TUBB3, TWNK, TYMP, VCP, VRK1, WARS, WNK1, YARS
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):	unknown
CREATED:	18 Jul 2019 - 10:28
CHANGED:	13 Oct 2021 - 15:46
URL:	https://laboboeken.nexuzhealth.com/pboek/internet/GHB%20CME/15337

RELATED CONTENT

Related Diseases

- ATTRV122I amyloidosis
- ATTRV30M amyloidosis
- Amyotrophic lateral sclerosis
- Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation
- Autosomal dominant Charcot-Marie-Tooth disease type 2DD
- Autosomal dominant Charcot-Marie-Tooth disease type 2E
- Autosomal dominant Charcot-Marie-Tooth disease type 2F
- Autosomal dominant Charcot-Marie-Tooth disease type 2I
- Autosomal dominant Charcot-Marie-Tooth disease type 2J
- Autosomal dominant Charcot-Marie-Tooth disease type 2K
- Autosomal dominant Charcot-Marie-Tooth disease type 2M
- Autosomal dominant Charcot-Marie-Tooth disease type 2N
- Autosomal dominant Charcot-Marie-Tooth disease type 2O
- Autosomal dominant Charcot-Marie-Tooth disease type 2V
- Autosomal dominant Charcot-Marie-Tooth disease type 2W
- Autosomal dominant Charcot-Marie-Tooth disease type 2Y
- Autosomal dominant intermediate Charcot-Marie-Tooth disease type B
- Autosomal dominant intermediate Charcot-Marie-Tooth disease type C
- Autosomal dominant intermediate Charcot-Marie-Tooth disease type D
- Autosomal dominant progressive external ophthalmoplegia
- Autosomal dominant slowed nerve conduction velocity
- Autosomal dominant spastic paraplegia type 17
- Autosomal dominant spastic paraplegia type 3
- Autosomal recessive Charcot-Marie-Tooth disease with hoarseness
- Autosomal recessive axonal neuropathy with neuromyotonia
- Autosomal recessive intermediate Charcot-Marie-Tooth disease type A

- Autosomal recessive intermediate Charcot-Marie-Tooth disease type B
- Autosomal recessive progressive external ophthalmoplegia
- Autosomal recessive spastic paraplegia type 55
- Autosomal spastic paraplegia type 30
- CADDS
- Charcot-Marie-Tooth disease type 1A
- Charcot-Marie-Tooth disease type 1B
- Charcot-Marie-Tooth disease type 1D
- Charcot-Marie-Tooth disease type 1E
- Charcot-Marie-Tooth disease type 1F
- Charcot-Marie-Tooth disease type 2B2
- Charcot-Marie-Tooth disease type 2B5
- Charcot-Marie-Tooth disease type 2H
- Charcot-Marie-Tooth disease type 4A
- Charcot-Marie-Tooth disease type 4B3
- Charcot-Marie-Tooth disease type 4E
- Charcot-Marie-Tooth disease type 4H
- DYNC1H1-related autosomal dominant childhood-onset proximal spinal muscular atrophy
- Dejerine-Sottas syndrome
- Distal hereditary motor neuropathy type 2
- Distal hereditary motor neuropathy type 5
- Fabry disease
- Giant axonal neuropathy
- Hereditary neuropathy with liability to pressure palsies
- Hereditary sensorimotor neuropathy with hyperelastic skin
- Hereditary sensory and autonomic neuropathy type 1
- Hereditary sensory and autonomic neuropathy type 2
- Hereditary sensory and autonomic neuropathy type 6
- Hereditary sensory and autonomic neuropathy type 8
- Hypomyelination neuropathy-arthrogryposis syndrome
- Muscular dystrophy, Selcen type
- Mutilating hereditary sensory neuropathy with spastic paraplegia
- Neuralgic amyotrophy

- Neuropathy with hearing impairment
- Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome
- Perrault syndrome
- Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome
- Pontocerebellar hypoplasia type 1
- SURF1-related Charcot-Marie-Tooth disease type 4
- X-linked Charcot-Marie-Tooth disease type 1
- X-linked Charcot-Marie-Tooth disease type 6

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- alanyl-tRNA synthetase 1
- ATP binding cassette subfamily D member 1
- abhydrolase domain containing 12, lysophospholipase
- ATP/GTP binding carboxypeptidase 1
- apoptosis inducing factor mitochondria associated 1
- aprataxin
- Rho guanine nucleotide exchange factor 10
- atlastin GTPase 1
- atlastin GTPase 3
- ATPase Na+/K+ transporting subunit alpha 1
- ATPase copper transporting alpha
- BAG cochaperone 3
- BICD cargo adaptor 2
- BSCL2 lipid droplet biogenesis associated, seipin
- chaperonin containing TCP1 subunit 5

- CD59 molecule (CD59 blood group)
- contactin associated protein 1
- cytochrome c oxidase assembly factor 7
- cytochrome c oxidase subunit 6A1
- CTD phosphatase subunit 1
- DDB1 and CUL4 associated factor 8
- dynactin subunit 1
- diacylglycerol O-acyltransferase 2
- dehydrogenase E1 and transketolase domain containing 1
- DnaJ heat shock protein family (Hsp40) member B2
- dynamin 2
- DNA methyltransferase 1
- dystrophin related protein 2
- dystonin
- dynein cytoplasmic 1 heavy chain 1
- early growth response 2
- elongator complex protein 1
- fibulin 5
- F-box protein 38
- FYVE, RhoGEF and PH domain containing 4
- FIG4 phosphoinositide 5-phosphatase
- FLVCR heme transporter 1
- gigaxonin
- glycyl-tRNA synthetase 1
- ganglioside induced differentiation associated protein 1
- gap junction protein beta 1
- gap junction protein beta 3
- galactosidase alpha
- G protein subunit beta 4
- hydroxyacyl-CoA dehydrogenase trifunctional multienzyme complex subunit alpha
- hydroxyacyl-CoA dehydrogenase trifunctional multienzyme complex subunit beta
- histidyl-tRNA synthetase 1
- histidine triad nucleotide binding protein 1

- hexokinase 1
- homeobox D10
- heat shock protein family B (small) member 1
- heat shock protein family B (small) member 3
- heat shock protein family B (small) member 8
- isoleucyl-tRNA synthetase 2, mitochondrial
- interferon related developmental regulator 1
- immunoglobulin mu DNA binding protein 2
- inverted formin, FH2 and WH2 domain containing
- inositol 1,4,5-trisphosphate receptor type 3
- lysyl-tRNA synthetase 1
- kinesin family member 1A
- kinesin family member 1B
- kinesin family member 5A
- kelch like family member 13
- lipopolysaccharide induced TNF factor
- lamin A/C
- leucine rich repeat and sterile alpha motif containing 1
- methionyl-tRNA synthetase 1
- minichromosome maintenance complex component 3 associated protein
- mediator complex subunit 12
- mediator complex subunit 25
- mitofusin 2
- membrane metalloendopeptidase
- MORC family CW-type zinc finger 2
- mitochondrial inner membrane protein MPV17
- myelin protein zero
- myotubularin related protein 2
- mitochondrial translation release factor in rescue
- myosin heavy chain 7B
- N-acetyl-alpha-glucosaminidase
- N-myc downstream regulated 1
- neurofilament heavy chain

- neurofilament light chain
- nerve growth factor
- neurotrophic receptor tyrosine kinase 1
- outer mitochondrial membrane lipid metabolism regulator OPA3
- pyruvate dehydrogenase kinase 3
- pyridoxal kinase
- peroxisomal biogenesis factor 1
- peroxisomal biogenesis factor 7
- phytanoyl-CoA 2-hydroxylase
- pleckstrin homology and RhoGEF domain containing G5
- peripheral myelin protein 2
- peripheral myelin protein 22
- polynucleotide kinase 3'-phosphatase
- DNA polymerase gamma, catalytic subunit
- PR/SET domain 12
- prion protein
- phosphoribosyl pyrophosphate synthetase 1
- periaxin
- RAB7A, member RAS oncogene family
- receptor accessory protein 1
- reticulophagy regulator 1
- SET binding factor 1
- SET binding factor 2
- sodium voltage-gated channel alpha subunit 10
- sodium voltage-gated channel alpha subunit 11
- sodium voltage-gated channel alpha subunit 9
- synthesis of cytochrome C oxidase 2
- sterol carrier protein 2
- septin 9
- senataxin
- sphingosine-1-phosphate lyase 1
- SH3 domain binding protein 4
- SH3 domain and tetratricopeptide repeats 2

- sigma non-opioid intracellular receptor 1
- solute carrier family 12 member 6
- solute carrier family 25 member 46
- solute carrier family 5 member 7
- sorbitol dehydrogenase
- SRY-box transcription factor 10
- SPG11 vesicle trafficking associated, spatacsin
- serine palmitoyltransferase long chain base subunit 1
- serine palmitoyltransferase long chain base subunit 2
- SURF1 cytochrome c oxidase assembly factor
- tyrosyl-DNA phosphodiesterase 1
- tectonin beta-propeller repeat containing 2
- trafficking from ER to golgi regulator
- tripartite motif containing 2
- transient receptor potential cation channel subfamily A member 1
- transient receptor potential cation channel subfamily V member 4
- transthyretin
- tubulin beta 3 class III
- twinkle mtDNA helicase
- thymidine phosphorylase
- valosin containing protein
- VRK serine/threonine kinase 1
- tryptophanyl-tRNA synthetase 1
- WNK lysine deficient protein kinase 1
- tyrosyl-tRNA synthetase 1

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
