
GENETIC TEST:
Leukodystrophy (gene panel)

FULL NAME:	Leukodystrophy (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:	30 Jul 2019 - 13:33
CHANGED:	16 Dec 2022 - 11:27

RELATED CONTENT

Related Laboratories

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

Related Analytes

- [alanyl-tRNA synthetase 2, mitochondrial](#)
- [4-aminobutyrate aminotransferase](#)
- [ATP binding cassette subfamily D member 1](#)
- [acyl-CoA binding domain containing 5](#)
- [ACD shelterin complex subunit and telomerase recruitment factor](#)
- [alkaline ceramidase 3](#)
- [acyl-CoA oxidase 1](#)
- [acid phosphatase 5, tartrate resistant](#)
- [actin like 6B](#)
- [adenosine deaminase RNA specific](#)
- [adhesion G protein-coupled receptor G1](#)
- [adenylosuccinate lyase](#)
- [aspartylglucosaminidase](#)
- [apoptosis inducing factor mitochondria associated 1](#)
- [aminoacyl tRNA synthetase complex interacting multifunctional protein 1](#)
- [aminoacyl tRNA synthetase complex interacting multifunctional protein 2](#)
- [aldehyde dehydrogenase 3 family member A2](#)
- [aldehyde dehydrogenase 5 family member A1](#)
- [amyloid beta precursor protein](#)
- [arylsulfatase A](#)

- N-acylsphingosine amidohydrolase 1
- aspartoacylase
- ATPase family AAA domain containing 3A
- ATPase copper transporting alpha
- ATPase copper transporting beta
- AU RNA binding methylglutaconyl-CoA hydratase
- beta-1,3-N-acetylgalactosaminyltransferase 2
- B cell receptor associated protein 31
- BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone
- bolA family member 3
- biotinidase
- cystathionine beta-synthase
- complement factor H
- capicua transcriptional repressor
- chloride voltage-gated channel 2
- CLN3 lysosomal/endosomal transmembrane protein, battenin
- CLN5 intracellular trafficking protein
- CLN6 transmembrane ER protein
- CLN8 transmembrane ER and ERGIC protein
- 2',3'-cyclic nucleotide 3' phosphodiesterase
- contactin associated protein 1
- cytochrome c oxidase assembly factor 8
- collagen type IV alpha 1 chain
- collagen type IV alpha 2 chain
- cytochrome c oxidase assembly factor heme A:farnesyltransferase COX10
- cytochrome c oxidase assembly homolog COX15
- colony stimulating factor 1 receptor
- CST telomere replication complex component 1
- cathepsin A
- cathepsin D
- cathepsin F
- cytochrome P450 family 21 subfamily A member 2
- cytochrome P450 family 27 subfamily A member 1

- dystroglycan 1
- aspartyl-tRNA synthetase 1
- aspartyl-tRNA synthetase 2, mitochondrial
- DDB1 and CUL4 associated factor 17
- DDHD domain containing 2
- delta 4-desaturase, sphingolipid 1
- deoxyguanosine kinase
- dihydrolipoamide S-acetyltransferase
- dihydrolipoamide dehydrogenase
- Dmx like 2
- DnaJ heat shock protein family (Hsp40) member C5
- glutamyl-tRNA synthetase 2, mitochondrial
- enoyl-CoA hydratase, short chain 1
- eukaryotic translation initiation factor 2 alpha kinase 1
- eukaryotic translation initiation factor 2 alpha kinase 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
- glutamyl-prolyl-tRNA synthetase 1
- ERCC excision repair 2, TFIIH core complex helicase subunit
- ERCC excision repair 3, TFIIH core complex helicase subunit
- ERCC excision repair 6, chromatin remodeling factor
- ERCC excision repair 8, CSA ubiquitin ligase complex subunit
- fatty acid 2-hydroxylase
- fatty acyl-CoA reductase 1
- phenylalanyl-tRNA synthetase 2, mitochondrial
- FAST kinase domains 2
- F-box and leucine rich repeat protein 4
- ferredoxin 2
- FIG4 phosphoinositide 5-phosphatase
- fukutin

- FLVCR heme transporter 2
- folate receptor 1
- alpha-L-fucosidase 1
- galactosylceramidase
- galactose-1-phosphate uridylyltransferase
- gigaxonin
- 1,4-alpha-glucan branching enzyme 1
- glial fibrillary acidic protein
- G elongation factor mitochondrial 1
- GTP dependent ribosome recycling factor mitochondrial 2
- gap junction protein alpha 1
- gap junction protein beta 1
- gap junction protein gamma 2
- galactosidase alpha
- galactosidase beta 1
- ganglioside GM2 activator
- GNAS complex locus
- gephyrin
- granulin precursor
- general transcription factor IIH subunit 5
- hepatic and glial cell adhesion molecule
- hexosaminidase subunit alpha
- hexosaminidase subunit beta
- heat shock protein nuclear import factor hiki
- holocarboxylase synthetase
- 3-hydroxy-3-methylglutaryl-CoA lyase
- hydroxysteroid 17-beta dehydrogenase 4
- heat shock protein family D (Hsp60) member 1
- HtrA serine peptidase 1
- hyccin PI4KA lipid kinase complex subunit 1
- iron-sulfur cluster assembly factor IBA57
- interferon induced with helicase C domain 1
- inhibitor of nuclear factor kappa B kinase regulatory subunit gamma

- iron-sulfur cluster assembly 2
- isovaleryl-CoA dehydrogenase
- junctional adhesion molecule 3
- lysyl-tRNA synthetase 1
- L-2-hydroxyglutarate dehydrogenase
- laminin subunit alpha 2
- LARGE xylosyl- and glucuronyltransferase 1
- leucyl-tRNA synthetase 2, mitochondrial
- lipoyltransferase 1
- lipoyl(octanoyl) transferase 2
- lamin B1
- LYR motif containing 7
- mannosidase alpha class 2B member 1
- mannosidase beta
- methionyl-tRNA synthetase 2, mitochondrial
- mucolipin TRP cation channel 1
- major facilitator superfamily domain containing 8
- modulator of VRAC current 1
- methylmalonyl-CoA mutase
- molybdenum cofactor synthesis 1
- molybdenum cofactor synthesis 2
- M-phase specific PLK1 interacting protein
- mitochondrial inner membrane protein MPV17
- mitochondrial ribosomal protein S34
- mitochondrial methionyl-tRNA formyltransferase
- methylenetetrahydrofolate reductase
- mitochondrial tRNA translation optimization 1
- NAD(P)HX dehydratase
- NAD(P)HX epimerase
- NADH:ubiquinone oxidoreductase subunit A2
- NADH:ubiquinone oxidoreductase subunit A9
- NADH:ubiquinone oxidoreductase complex assembly factor 1
- NADH:ubiquinone oxidoreductase complex assembly factor 3

- 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 S2
- NADH:ubiquinone oxidoreductase subunit S4
- NADH:ubiquinone oxidoreductase core subunit S7
- NADH:ubiquinone oxidoreductase core subunit S8
- NADH:ubiquinone oxidoreductase core subunit V1
- neuraminidase 1
- NK6 homeobox 2
- notch receptor 3
- NPC intracellular cholesterol transporter 1
- NPC intracellular cholesterol transporter 2
- netrin G2
- NUBP iron-sulfur cluster assembly factor, mitochondrial
- occludin
- OCRL inositol polyphosphate-5-phosphatase
- phenylalanine hydroxylase
- prolyl-tRNA synthetase 2, mitochondrial
- propionyl-CoA carboxylase subunit alpha
- propionyl-CoA carboxylase subunit beta
- platelet derived growth factor subunit B
- platelet derived growth factor receptor beta
- pyruvate dehydrogenase E1 subunit alpha 1
- pyruvate dehydrogenase E1 subunit beta
- pyruvate dehydrogenase complex component X
- pyruvate dehydrogenase phosphatase catalytic subunit 1
- peroxisomal biogenesis factor 1
- peroxisomal biogenesis factor 10
- peroxisomal biogenesis factor 11 beta
- peroxisomal biogenesis factor 12
- peroxisomal biogenesis factor 13
- peroxisomal biogenesis factor 14

- peroxisomal biogenesis factor 16
- peroxisomal biogenesis factor 19
- peroxisomal biogenesis factor 2
- peroxisomal biogenesis factor 26
- peroxisomal biogenesis factor 3
- peroxisomal biogenesis factor 5
- peroxisomal biogenesis factor 6
- peroxisomal biogenesis factor 7
- pleckstrin homology and RhoGEF domain containing G2
- proteolipid protein 1
- pyridoxal phosphate binding protein
- peptidase, mitochondrial processing subunit beta
- polyribonucleotide nucleotidyltransferase 1
- DNA polymerase gamma, catalytic subunit
- RNA polymerase I and III subunit C
- RNA polymerase III subunit A
- RNA polymerase III subunit B
- RNA polymerase III subunit K
- protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-)
- protein O-mannosyltransferase 1
- protein O-mannosyltransferase 2
- palmitoyl-protein thioesterase 1
- perforin 1
- prosaposin
- phosphoserine aminotransferase 1
- phosphoserine phosphatase
- pyrroline-5-carboxylate reductase 2
- quinoid dihydropteridine reductase
- arginyl-tRNA synthetase 1
- ribonuclease H2 subunit A
- ribonuclease H2 subunit B
- ribonuclease H2 subunit C
- ribonuclease T2

- SAM and HD domain containing deoxynucleoside triphosphate triphosphohydrolase 1
- sterol carrier protein 2
- succinate dehydrogenase complex flavoprotein subunit A
- succinate dehydrogenase complex assembly factor 1
- succinate dehydrogenase complex assembly factor 2
- succinate dehydrogenase complex iron sulfur subunit B
- succinate dehydrogenase complex subunit D
- Sep (O-phosphoserine) tRNA:Sec (selenocysteine) tRNA synthase
- shugoshin 1
- SH3 and multiple ankyrin repeat domains 3
- solute carrier family 13 member 3
- solute carrier family 16 member 2
- solute carrier family 17 member 5
- solute carrier family 20 member 2
- solute carrier family 25 member 12
- solute carrier family 44 member 1
- solute carrier family 7 member 2
- synaptosome associated protein 29
- small nucleolar RNA, C/D box 118
- SRY-box transcription factor 10
- SPG11 vesicle trafficking associated, spatacsin
- STN1 subunit of CST complex
- syntaxin 11
- syntaxin binding protein 2
- succinate-CoA ligase ADP-forming subunit beta
- sulfatase modifying factor 1
- sulfite oxidase
- SURF1 cytochrome c oxidase assembly factor
- translational activator of cytochrome c oxidase I
- transmembrane protein 106B
- transmembrane protein 63A
- triggering receptor expressed on myeloid cells 2
- three prime repair exonuclease 1

- tRNA splicing endonuclease subunit 54
- transthyretin
- tubulin beta 4A class IVa
- twinkle mtDNA helicase
- thymidine phosphorylase
- transmembrane immune signaling adaptor TYROBP
- upstream binding transcription factor
- ubiquitin fold modifier 1
- unc-13 homolog D
- VPS11 core subunit of CORVET and HOPS complexes
- tryptophanyl tRNA synthetase 2, mitochondrial
- WT1 transcription factor
- xenotropic and polytropic retrovirus receptor 1
- zinc finger FYVE-type containing 26

Related Gene Panels

- Leukodystrophy - UGent

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