GENETIC TEST:
Epilepsy (gene panel)

FULL NAME:	Epilepsy (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Whole Exome Sequencing (WES)
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2023-11-09 / 2024-05-08
EQA:	• epilepsy disorders, • epilepsy disorders
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	29 Nov 2019 - 15:33

CHANGED:	22 Jan 2024 - 11:38
URL:	https://labogidsmedgen.uza.be/analyses/epilepsie-geassocieerd-met-koortsstuipen…

Source URL: http://gentest.healthdata.be/genetic_test/954

RELATED CONTENT

Related Diseases

- 17q11 microdeletion syndrome
- 9q33.3q34.11 microdeletion syndrome
- Action myoclonus-renal failure syndrome
- Atypical Rett syndrome
- Autosomal dominant epilepsy with auditory features
- Autosomal dominant nocturnal frontal lobe epilepsy
- Autosomal dominant non-syndromic intellectual disability
- Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency
- Autosomal recessive non-syndromic intellectual disability
- Baraitser-Winter cerebrofrontofacial syndrome
- Benign familial infantile epilepsy
- Benign familial neonatal epilepsy
- Benign familial neonatal-infantile seizures
- Bilateral generalized polymicrogyria
- CDKL5-deficiency disorder
- CLN1 disease
- CLN10 disease
- CLN2 disease
- CLN3 disease
- CLN4A disease
- CLN5 disease
- CLN6 disease
- CLN7 disease
- CLN8 disease
- CNTNAP2-related developmental and epileptic encephalopathy
- Childhood absence epilepsy

- Christianson syndrome
- Continuous spikes and waves during sleep
- Corpus callosum agenesis-abnormal genitalia syndrome
- Craniosynostosis-microretrognathia-severe intellectual disability syndrome
- Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome
- Dravet syndrome
- EAST syndrome
- Early infantile epileptic encephalopathy
- Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation
- Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome
- Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome
- Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome
- Epileptic encephalopathy with global cerebral demyelination
- FOXG1 syndrome
- Familial encephalopathy with neuroserpin inclusion bodies
- Familial focal epilepsy with variable foci
- Familial infantile myoclonic epilepsy
- Familial porencephaly
- Familial schizencephaly
- Female restricted epilepsy with intellectual disability
- Focal epilepsy-intellectual disability-cerebro-cerebellar malformation
- Generalized epilepsy with febrile seizures-plus
- Generalized epilepsy-paroxysmal dyskinesia syndrome
- Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome
- Hyperekplexia-epilepsy syndrome
- Infantile convulsions and choreoathetosis
- Infantile epileptic-dyskinetic encephalopathy
- Infantile spasms syndrome
- Isolated focal cortical dysplasia type IIa
- Isolated focal cortical dysplasia type IIb
- Isolated focal cortical dysplasia type Ia
- Juvenile myoclonic epilepsy
- KCNQ2-related epileptic encephalopathy

- KDM5C-related syndromic X-linked intellectual disability
- Kleefstra syndrome due to 9q34 microdeletion
- Lafora disease
- Landau-Kleffner syndrome
- Lennox-Gastaut syndrome
- Lissencephaly due to LIS1 mutation
- Lissencephaly due to TUBA1A mutation
- Lissencephaly syndrome, Norman-Roberts type
- Lissencephaly type 1 due to doublecortin gene mutation
- Malignant migrating focal seizures of infancy
- Miller-Dieker syndrome
- Mowat-Wilson syndrome due to a ZEB2 point mutation
- Mowat-Wilson syndrome due to monosomy 2q22
- Myoclonic-astatic epilepsy
- Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion
- Non-specific early-onset epileptic encephalopathy
- PEHO syndrome
- Partington syndrome
- Periventricular nodular heterotopia
- Pitt-Hopkins syndrome
- Pitt-Hopkins-like syndrome
- Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome
- Progressive epilepsy-intellectual disability syndrome, Finnish type
- Progressive myoclonic epilepsy type 1
- Progressive myoclonic epilepsy type 3
- Progressive myoclonic epilepsy type 5
- Progressive myoclonic epilepsy type 6
- Progressive myoclonic epilepsy type 7
- Progressive myoclonic epilepsy type 8
- Progressive myoclonic epilepsy with dystonia
- Proteus syndrome
- Rett syndrome
- Rolandic epilepsy

- Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome
- Rolandic epilepsy-speech dyspraxia syndrome
- SYNGAP1-related developmental and epileptic encephalopathy
- Semilobar holoprosencephaly
- Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome
- Severe neonatal-onset encephalopathy with microcephaly
- Thiamine-responsive encephalopathy
- Tuberous sclerosis complex
- Walker-Warburg syndrome
- X-linked epilepsy-learning disabilities-behavior disorders syndrome
- X-linked lissencephaly with abnormal genitalia
- X-linked non-syndromic intellectual disability
- X-linked spasticity-intellectual disability-epilepsy syndrome

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- alanyl-tRNA synthetase 1
- 4-aminobutyrate aminotransferase
- ACD shelterin complex subunit and telomerase recruitment factor
- actin beta
- actin gamma 1
- actin like 6B
- aminoacylase 1
- adenosine deaminase RNA specific
- ADP-ribosylserine hydrolase
- adenylosuccinate lyase

- AFG2 AAA ATPase homolog A
- AFG3 like matrix AAA peptidase subunit 2
- aldehyde dehydrogenase 7 family member A1
- ALG11 alpha-1,2-mannosyltransferase
- ALG13 UDP-N-acetylglucosaminyltransferase subunit
- aminomethyltransferase
- ankyrin repeat domain containing 11
- adaptor related protein complex 3 subunit beta 2
- adaptor related protein complex 4 subunit sigma 1
- ADP ribosylation factor guanine nucleotide exchange factor 2
- Cdc42 guanine nucleotide exchange factor 9
- AT-rich interaction domain 1B
- ARV1 homolog, fatty acid homeostasis modulator
- aristaless related homeobox
- ASXL transcriptional regulator 3
- atrophin 1
- ATPase Na+/K+ transporting subunit alpha 2
- ATPase Na+/K+ transporting subunit alpha 3
- BRCA1 associated ATM activator 1
- BSCL2 lipid droplet biogenesis associated, seipin
- calcium voltage-gated channel subunit alpha1 A
- calcium voltage-gated channel subunit alpha1 B
- calcium voltage-gated channel subunit alpha1 E
- calcium voltage-gated channel subunit alpha1 G
- calcium voltage-gated channel auxiliary subunit alpha2delta 2
- calcium/calmodulin dependent serine protein kinase
- cyclin dependent kinase 13
- cyclin dependent kinase like 5
- ceramide synthase 1
- ceramide transporter 1
- chromodomain helicase DNA binding protein 2
- chloride voltage-gated channel 4
- CLN3 lysosomal/endosomal transmembrane protein, battenin

- CLN5 intracellular trafficking protein
- CLN6 transmembrane ER protein
- CLN8 transmembrane ER and ERGIC protein
- clathrin heavy chain
- connector enhancer of kinase suppressor of Ras 2
- contactin associated protein 2
- collagen type XVIII alpha 1 chain
- collagen type IV alpha 1 chain
- complexin 1
- carnitine palmitoyltransferase 2
- casein kinase 2 beta
- cystatin B
- cathepsin D
- cut like homeobox 2
- cytoplasmic FMR1 interacting protein 2
- doublecortin
- DEAD-box helicase 3 X-linked
- DENN domain containing 5A
- DEP domain containing 5, GATOR1 subcomplex subunit
- dehydrodolichyl diphosphate synthase subunit
- Dmx like 2
- dynamin 1
- dynamin 1 like
- dedicator of cytokinesis 7
- dual specificity tyrosine phosphorylation regulated kinase 1A
- eukaryotic translation elongation factor 1 alpha 2
- euchromatic histone lysine methyltransferase 1
- EPM2A glucan phosphatase, laforin
- phenylalanyl-tRNA synthetase 2, mitochondrial
- fibroblast growth factor 12
- filamin A
- formin 2
- fragile X messenger ribonucleoprotein 1

- folate receptor 1
- forkhead box G1
- ferric chelate reductase 1 like
- fizzy and cell division cycle 20 related 1
- gamma-aminobutyric acid type B receptor subunit 2
- gamma-aminobutyric acid type A receptor subunit alpha1
- gamma-aminobutyric acid type A receptor subunit alpha2
- gamma-aminobutyric acid type A receptor subunit beta1
- gamma-aminobutyric acid type A receptor subunit beta2
- gamma-aminobutyric acid type A receptor subunit beta3
- gamma-aminobutyric acid type A receptor subunit gamma2
- glutamate decarboxylase 1
- guanidinoacetate N-methyltransferase
- glucosylceramidase beta 1
- glycine decarboxylase
- G protein subunit alpha o1
- G protein subunit beta 1
- G protein subunit beta 5
- golgi SNAP receptor complex member 2
- gephyrin
- glutamate ionotropic receptor AMPA type subunit 2
- glutamate ionotropic receptor AMPA type subunit 3
- glutamate ionotropic receptor kainate type subunit 2
- glutamate ionotropic receptor NMDA type subunit 1
- glutamate ionotropic receptor NMDA type subunit 2A
- glutamate ionotropic receptor NMDA type subunit 2B
- glutamate ionotropic receptor NMDA type subunit 2D
- hyperpolarization activated cyclic nucleotide gated potassium channel 1
- HECT, C2 and WW domain containing E3 ubiquitin protein ligase 2
- heterogeneous nuclear ribonucleoprotein U
- HECT, UBA and WWE domain containing E3 ubiquitin protein ligase 1
- immediate early response 3 interacting protein 1
- interferon induced with helicase C domain 1

- IQ motif and Sec7 domain ArfGEF 2
- interferon regulatory factor 2 binding protein like
- inosine triphosphatase
- KAT8 regulatory NSL complex subunit 1
- lysyl-tRNA synthetase 1
- potassium voltage-gated channel subfamily A member 1
- potassium voltage-gated channel subfamily A member 2
- potassium voltage-gated channel subfamily B member 1
- potassium voltage-gated channel subfamily C member 1
- potassium voltage-gated channel subfamily H member 1
- potassium inwardly rectifying channel subfamily J member 10
- potassium two pore domain channel subfamily K member 4
- potassium calcium-activated channel subfamily M alpha 1
- potassium voltage-gated channel subfamily Q member 2
- potassium voltage-gated channel subfamily Q member 3
- potassium voltage-gated channel subfamily Q member 5
- potassium sodium-activated channel subfamily T member 1
- potassium sodium-activated channel subfamily T member 2
- potassium channel tetramerization domain containing 7
- lysine demethylase 5C
- kinesin family member 1A
- kinesin family member 2A
- kinesin family member 5C
- lysine methyltransferase 2E (inactive)
- microtubule associated protein 1B
- methyl-CpG binding domain protein 5
- methyl-CpG binding protein 2
- mediator complex subunit 13L
- myocyte enhancer factor 2C
- major facilitator superfamily domain containing 8
- molybdenum cofactor synthesis 1
- molybdenum cofactor synthesis 2
- mechanistic target of rapamycin kinase

- asparaginyl-tRNA synthetase 2, mitochondrial
- neurobeachin
- neuraminidase 1
- neurite extension and migration factor
- neurofibromin 1
- NF2, moesin-ezrin-radixin like (MERLIN) tumor suppressor
- NHL repeat containing E3 ubiquitin protein ligase 1
- NIPA magnesium transporter 1
- NPR2 like, GATOR1 complex subunit
- NPR3 like, GATOR1 complex subunit
- neurexin 1
- neurotrophic receptor tyrosine kinase 2
- NUS1 dehydrodolichyl diphosphate synthase subunit
- OTU deubiquitinase 6B
- phosphofurin acidic cluster sorting protein 2
- platelet activating factor acetylhydrolase 1b regulatory subunit 1
- prolyl-tRNA synthetase 2, mitochondrial
- protocadherin 19
- phosphatase and actin regulator 1
- phosphatidylinositol glycan anchor biosynthesis class A
- phosphatidylinositol glycan anchor biosynthesis class N
- phosphatidylinositol glycan anchor biosynthesis class Q
- phosphatidylinositol glycan anchor biosynthesis class S
- phosphatidylinositol glycan anchor biosynthesis class T
- phospholipase C beta 1
- pyridoxal phosphate binding protein
- polynucleotide kinase 3'-phosphatase
- pyridoxamine 5'-phosphate oxidase
- DNA polymerase gamma, catalytic subunit
- protein phosphatase 2 regulatory subunit B'delta
- protein phosphatase 3 catalytic subunit alpha
- palmitoyl-protein thioesterase 1
- prickle planar cell polarity protein 1

- proline rich transmembrane protein 2
- phosphatase and tensin homolog
- purine rich element binding protein A
- glutaminyl-tRNA synthetase 1
- reelin
- Rho related BTB domain containing 2
- ribonuclease H2 subunit A
- ribonuclease H2 subunit B
- ribonuclease H2 subunit C
- rogdi atypical leucine zipper
- RAR related orphan receptor A
- RAR related orphan receptor B
- reticulon 4 interacting protein 1
- SAM and HD domain containing deoxynucleoside triphosphate triphosphohydrolase 1
- scavenger receptor class B member 2
- sodium voltage-gated channel alpha subunit 1
- sodium voltage-gated channel beta subunit 1
- sodium voltage-gated channel alpha subunit 2
- sodium voltage-gated channel alpha subunit 3
- sodium voltage-gated channel alpha subunit 8
- serpin family I member 1
- SET domain containing 2, histone lysine methyltransferase
- salt inducible kinase 1
- solute carrier family 12 member 5
- solute carrier family 13 member 5
- solute carrier family 19 member 3
- solute carrier family 1 member 2
- solute carrier family 25 member 12
- solute carrier family 25 member 22
- solute carrier family 2 member 1
- solute carrier family 35 member A2
- solute carrier family 35 member A3
- solute carrier family 6 member 1

- solute carrier family 6 member 8
- solute carrier family 9 member A6
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 2
- structural maintenance of chromosomes 1A
- synaptosome associated protein 25
- spectrin alpha, non-erythrocytic 1
- ST3 beta-galactoside alpha-2,3-sialyltransferase 3
- ST3 beta-galactoside alpha-2,3-sialyltransferase 5
- STAG1 cohesin complex component
- syntaxin 1B
- syntaxin binding protein 1
- synapsin I
- synaptic Ras GTPase activating protein 1
- synaptojanin 1
- SZT2 subunit of KICSTOR complex
- tetratricopeptide repeat, ankyrin repeat and coiled-coil containing 2
- TBC1 domain family member 24
- tubulin folding cofactor D
- transcription factor 4
- thymidine kinase 2
- trafficking kinesin protein 1
- trafficking protein particle complex 6B
- three prime repair exonuclease 1
- tRNA isopentenyltransferase 1
- TSC complex subunit 1
- TSC complex subunit 2
- tubulin alpha 1a
- tubulin beta 2B class IIb
- ubiquitin like modifier activating enzyme 5
- ubiquitin protein ligase E3A
- ubiquitin-fold modifier conjugating enzyme 1
- UDP-glucose 6-dehydrogenase
- valyl-tRNA synthetase 1

- valyl-tRNA synthetase 2, mitochondrial
- WD repeat domain 26
- WD repeat domain 45
- WW domain containing oxidoreductase
- tyrosine 3-monooxygenase/tryptophan 5-monooxygenase activation protein gamma
- zinc finger and BTB domain containing 18
- zinc finger E-box binding homeobox 2
- zinc finger HIT-type containing 3

Related Gene Panels

- Rare epilepsy with developmental delay (> 240 genes) - UZA
