GENETIC TEST:
Lysosomal Storage Disease (gene panel)

FULL NAME:	Lysosomal Storage Disease (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal Diagnosis, Pre-implantation genetic diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Dried blood spot card
METHOD CATEGORY:	Sequence analysis: entire coding region Mutation screening and sequence analysis of selected exons
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-10-07 / 2026-06-14

EQA:	• next generation sequencing (germline), • next generation sequencing (germline), • next generation sequencing (germline), • next generation sequencing (germline) , • next generation sequencing (germline)
TURNAROUND TIME (MAXIMUM):	3 months
CREATED:	26 Aug 2019 - 12:39
CHANGED:	09 Mar 2023 - 16:03
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Lysosomale%20stapelingsziekte…

RELATED CONTENT

Related Diseases

- Adult Krabbe disease
- Chronic visceral acid sphingomyelinase deficiency
- GM1 gangliosidosis type 1
- GM1 gangliosidosis type 2
- GM1 gangliosidosis type 3
- Hurler-Scheie syndrome
- Infantile Krabbe disease
- Infantile neurovisceral acid sphingomyelinase deficiency
- Late-infantile/juvenile Krabbe disease
- Metachromatic leukodystrophy, adult form
- Metachromatic leukodystrophy, juvenile form
- Metachromatic leukodystrophy, late infantile form
- Mucolipidosis type II
- Mucolipidosis type III alpha/beta
- Mucolipidosis type III gamma
- Mucopolysaccharidosis type 2, attenuated form
- Mucopolysaccharidosis type 2, severe form
- Mucopolysaccharidosis type 4A
- Mucopolysaccharidosis type 4B
- Mucopolysaccharidosis type 7
- Niemann-Pick disease type C, adult neurologic onset
- Niemann-Pick disease type C, juvenile neurologic onset
- Niemann-Pick disease type C, late infantile neurologic onset
- Niemann-Pick disease type C, severe early infantile neurologic onset
- Niemann-Pick disease type C, severe perinatal form
- Sandhoff disease, adult form

- Sandhoff disease, infantile form
- Sandhoff disease, juvenile form
- Sanfilippo syndrome type A
- Sanfilippo syndrome type B
- Sialidosis type 1
- Tay-Sachs disease, B variant, adult form
- Tay-Sachs disease, B variant, infantile form
- Tay-Sachs disease, B variant, juvenile form
- Tay-Sachs disease, B1 variant

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- ACD shelterin complex subunit and telomerase recruitment factor
- adenylate cyclase 5
- aspartylglucosaminidase
- arylsulfatase A
- N-acylsphingosine amidohydrolase 1
- Chitinase 1
- CLN3 lysosomal/endosomal transmembrane protein, battenin
- CLN5 intracellular trafficking protein
- CLN6 transmembrane ER protein
- CLN8 transmembrane ER and ERGIC protein
- cystinosin, lysosomal cystine transporter
- cathepsin A
- cathepsin C
- cathepsin D

- cathepsin K
- deoxyguanosine kinase
- F-box and leucine rich repeat protein 4
- alpha-L-fucosidase 1
- alpha glucosidase
- galactosylceramidase
- galactosamine (N-acetyl)-6-sulfatase
- glucosylceramidase beta 1
- galactosidase alpha
- galactosidase beta 1
- ganglioside GM2 activator
- N-acetylglucosamine-1-phosphate transferase subunits alpha and beta
- N-acetylglucosamine-1-phosphate transferase subunit gamma
- glucosamine (N-acetyl)-6-sulfatase
- glucuronidase beta
- hexosaminidase subunit alpha
- hexosaminidase subunit beta
- heparan-alpha-glucosaminide N-acetyltransferase
- hyaluronidase 1
- iduronate 2-sulfatase
- alpha-L-iduronidase
- lysosomal associated membrane protein 2
- lipase A, lysosomal acid type
- mannosidase alpha class 2B member 1
- mannosidase beta
- mucolipin TRP cation channel 1
- major facilitator superfamily domain containing 8
- mitochondrial genome maintenance exonuclease 1
- mitochondrial inner membrane protein MPV17
- alpha-N-acetylgalactosaminidase
- N-acetyl-alpha-glucosaminidase
- neuraminidase 1
- NPC intracellular cholesterol transporter 1

- NPC intracellular cholesterol transporter 2
- OCRL inositol polyphosphate-5-phosphatase
- DNA polymerase gamma, catalytic subunit
- palmitoyl-protein thioesterase 1
- prosaposin
- ribonucleotide reductase regulatory TP53 inducible subunit M2B
- N-sulfoglucosamine sulfohydrolase
- solute carrier family 17 member 5
- secreted LY6/PLAUR domain containing 1
- sphingomyelin phosphodiesterase 1
- steroid sulfatase
- succinate-CoA ligase ADP-forming subunit beta
- succinate-CoA ligase GDP/ADP-forming subunit alpha
- sulfatase modifying factor 1
- thymidine kinase 2
- twinkle mtDNA helicase
- thymidine phosphorylase

Related Gene Panels

- Lysomal Storage (64 genes) - VUB
