GENETIC TEST:
Metabolic disorders including disorders of glycosylation, peroxisomal disorders, organic acidurias, glycogenosis disorders, neurotransmitter disorders (213 genes)

FULL NAME:	Metabolic disorders including disorders of glycosylation, peroxisomal disorders, organic acidurias, glycogenosis disorders, neurotransmitter disorders (213 genes)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal Diagnosis, Pre-implantation genetic diagnosis, Prenatal 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):	6 months (2 months for neonatal testing)
CREATED:	26 Aug 2019 - 14:50
CHANGED:	09 Mar 2023 - 16:08
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Metabole%20aandoening&&&&&287…

RELATED CONTENT

Related Diseases

- 3-hydroxy-3-methylglutaric aciduria
- Argininosuccinic aciduria
- Congenital disorder of glycosylation
- Glycogen storage disease due to glycogen debranching enzyme deficiency
- Hereditary orotic aciduria
- Malonic aciduria
- Methylmalonic aciduria due to transcobalamin receptor defect
- Mevalonic aciduria
- Oxoglutaric aciduria
- Peroxisomal acyl-CoA oxidase deficiency
- Urocanic aciduria
- Vitamin B12-responsive methylmalonic acidemia type cblB

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- 4-aminobutyrate aminotransferase
- ATP binding cassette subfamily D member 1
- acetyl-CoA acetyltransferase 1
- acyl-CoA oxidase 1

- amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase
- alkylglycerone phosphate synthase
- alanine--glyoxylate aminotransferase
- aldehyde dehydrogenase 18 family member A1
- aldehyde dehydrogenase 7 family member A1
- aldolase, fructose-bisphosphate A
- aldolase, fructose-bisphosphate B
- ALG1 chitobiosyldiphosphodolichol beta-mannosyltransferase
- ALG10 alpha-1,2-glucosyltransferase
- ALG11 alpha-1,2-mannosyltransferase
- ALG12 alpha-1,6-mannosyltransferase
- ALG13 UDP-N-acetylglucosaminyltransferase subunit
- ALG14 UDP-N-acetylglucosaminyltransferase subunit
- ALG2 alpha-1,3/1,6-mannosyltransferase
- ALG3 alpha-1,3- mannosyltransferase
- ALG5 dolichyl-phosphate beta-glucosyltransferase
- ALG6 alpha-1,3-glucosyltransferase
- ALG8 alpha-1,3-glucosyltransferase
- ALG9 alpha-1,2-mannosyltransferase
- alpha-methylacyl-CoA racemase
- aminomethyltransferase
- aspartoacylase
- ATPase H+ transporting accessory protein 1
- ATPase H+ transporting V0 subunit a2
- ATPase H+ transporting V1 subunit A
- ATPase H+ transporting V1 subunit E1
- ATPase copper transporting alpha
- ATPase phospholipid transporting 9B (putative)
- beta-1,3-N-acetylgalactosaminyltransferase 2
- beta-1,3-galactosyltransferase 6
- beta-1,3-glucuronyltransferase 3
- beta 3-glucosyltransferase
- beta-1,4-N-acetyl-galactosaminyltransferase 1

- beta-1,4-galactosyltransferase 1
- beta-1,4-galactosyltransferase 7
- branched chain keto acid dehydrogenase E1 subunit alpha
- branched chain keto acid dehydrogenase E1 subunit beta
- biotinidase
- carbamoyl-phosphate synthetase 2, aspartate transcarbamylase, and dihydroorotase
- coiled-coil domain containing 115
- carbohydrate sulfotransferase 14
- carbohydrate sulfotransferase 3
- carbohydrate sulfotransferase 6
- chondroitin sulfate synthase 1
- component of oligomeric golgi complex 1
- component of oligomeric golgi complex 2
- component of oligomeric golgi complex 3
- component of oligomeric golgi complex 4
- component of oligomeric golgi complex 5
- component of oligomeric golgi complex 6
- component of oligomeric golgi complex 7
- component of oligomeric golgi complex 8
- CDP-L-ribitol pyrophosphorylase A
- cystathionine gamma-lyase
- D-2-hydroxyglutarate dehydrogenase
- defender against cell death 1
- dopamine beta-hydroxylase
- dihydrolipoamide branched chain transacylase E2
- dopa decarboxylase
- dolichyl-diphosphooligosaccharide--protein glycosyltransferase non-catalytic subunit
- dehydrodolichyl diphosphate synthase subunit
- dynamin 1 like
- dolichol kinase
- dolichyl-phosphate N-acetylglucosaminephosphotransferase 1
- dolichyl-phosphate mannosyltransferase subunit 1, catalytic
- dolichyl-phosphate mannosyltransferase subunit 2, regulatory

- dolichyl-phosphate mannosyltransferase subunit 3, regulatory
- enolase 3
- EGF domain specific O-linked N-acetylglucosamine transferase
- exostosin glycosyltransferase 1
- exostosin glycosyltransferase 2
- FA complementation group A
- fukutin related protein
- fukutin
- fucosyltransferase 1 (H blood group)
- glucose-6-phosphatase catalytic subunit 1
- alpha glucosidase
- gamma-aminobutyric acid type A receptor subunit gamma2
- UDP-galactose-4-epimerase
- galactokinase 1
- polypeptide N-acetylgalactosaminyltransferase 3
- galactose-1-phosphate uridylyltransferase
- guanidinoacetate N-methyltransferase
- glucosidase II alpha subunit
- glycine amidinotransferase
- 1,4-alpha-glucan branching enzyme 1
- glutaryl-CoA dehydrogenase
- GTP cyclohydrolase 1
- glycine cleavage system protein H
- glutamine--fructose-6-phosphate transaminase 1
- glycine decarboxylase
- GDP-mannose pyrophosphorylase A
- GDP-mannose pyrophosphorylase B
- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase
- glycine N-methyltransferase
- glyceronephosphate O-acyltransferase
- glycogen synthase 1
- glycogen synthase 2
- homogentisate 1,2-dioxygenase

- holocarboxylase synthetase
- 3-hydroxy-3-methylglutaryl-CoA lyase
- hypoxanthine phosphoribosyltransferase 1
- hydroxysteroid 17-beta dehydrogenase 4
- isovaleryl-CoA dehydrogenase
- lysosomal associated membrane protein 2
- LARGE xylosyl- and glucuronyltransferase 1
- lactate dehydrogenase A
- LFNG O-fucosylpeptide 3-beta-N-acetylglucosaminyltransferase
- magnesium transporter 1
- mannosidase alpha class 1B member 1
- monoamine oxidase A
- methylcrotonyl-CoA carboxylase subunit 1
- methylcrotonyl-CoA carboxylase subunit 2
- alpha-1,6-mannosyl-glycoprotein 2-beta-N-acetylglucosaminyltransferase
- metabolism of cobalamin associated A
- metabolism of cobalamin associated B
- metabolism of cobalamin associated C
- methylmalonyl-CoA mutase
- mannosyl-oligosaccharide glucosidase
- mannose-P-dolichol utilization defect 1
- mannose phosphate isomerase
- N-acetylneuraminate synthase
- N-glycanase 1
- NUS1 dehydrodolichyl diphosphate synthase subunit
- ornithine aminotransferase
- oligosaccharyltransferase complex subunit 4, non-catalytic
- propionyl-CoA carboxylase subunit alpha
- propionyl-CoA carboxylase subunit beta
- 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
- phosphofructokinase, muscle
- phosphoglycerate mutase 2
- post-GPI attachment to proteins inositol deacylase 1
- post-GPI attachment to proteins 2
- post-GPI attachment to proteins phospholipase 3
- phosphoglucomutase 1
- phosphoglucomutase 2
- phosphoglucomutase 3
- phosphoglycerate dehydrogenase
- phosphorylase kinase regulatory subunit alpha 1
- phosphorylase kinase regulatory subunit alpha 2
- phosphorylase kinase regulatory subunit beta
- phosphorylase kinase catalytic subunit gamma 2
- phosphatidylinositol glycan anchor biosynthesis class A
- phosphatidylinositol glycan anchor biosynthesis class C
- phosphatidylinositol glycan anchor biosynthesis class G
- phosphatidylinositol glycan anchor biosynthesis class L
- phosphatidylinositol glycan anchor biosynthesis class M
- phosphatidylinositol glycan anchor biosynthesis class N
- phosphatidylinositol glycan anchor biosynthesis class O
- phosphatidylinositol glycan anchor biosynthesis class V
- phosphatidylinositol glycan anchor biosynthesis class W
- phosphatidylinositol glycan anchor biosynthesis class Y

- phosphomannomutase 2
- pyridoxamine 5'-phosphate oxidase
- protein O-fucosyltransferase 1
- protein O-glucosyltransferase 1
- protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-)
- protein O-mannosyltransferase 1
- protein O-mannosyltransferase 2
- protein kinase C substrate 80K-H
- phosphoserine aminotransferase 1
- glycogen phosphorylase L
- glycogen phosphorylase, muscle associated
- RFT1 homolog
- ribophorin I
- ribophorin II
- ribitol xylosyltransferase 1
- sterol carrier protein 2
- SEC23 homolog B, COPII coat complex component
- solute carrier family 25 member 22
- solute carrier family 2 member 1
- solute carrier family 35 member A1
- solute carrier family 35 member A2
- solute carrier family 35 member A3
- solute carrier family 35 member C1
- solute carrier family 35 member D1
- solute carrier family 39 member 8
- solute carrier family 3 member 1
- solute carrier family 6 member 19
- solute carrier family 6 member 8
- solute carrier family 7 member 9
- sepiapterin reductase
- steroid 5 alpha-reductase 3
- signal sequence receptor subunit 3
- signal sequence receptor subunit 4

- ST3 beta-galactoside alpha-2,3-sialyltransferase 3
- ST3 beta-galactoside alpha-2,3-sialyltransferase 5
- STT3 oligosaccharyltransferase complex catalytic subunit A
- STT3 oligosaccharyltransferase complex catalytic subunit B
- transmembrane protein 165
- transmembrane protein 199
- trafficking protein particle complex subunit 11
- tumor suppressor candidate 3
- vacuolar protein sorting 13 homolog B
- xylosyltransferase 1
- xylosyltransferase 2

Related Gene Panels

- Metabolic disorders (213 genes) - VUB
