GENETIC TEST:
Metabolic diseases with hepatic disorders (20 genes)

FULL NAME:	Metabolic diseases with hepatic disorders (20 genes)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Post-natal Diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Amniotic fluid, Chorionic villi
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2022-12-22 / 2027-12-21
TURNAROUND TIME (MAXIMUM):	3 months

CREATED:	06 Nov 2019 - 14:22
CHANGED:	25 Jan 2023 - 08:38

Source URL: http://gentest.healthdata.be/genetic_test/926

RELATED CONTENT

Related Diseases

- Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins
- CPT deficiency, hepatic, type IA
- Carnitine palmitoyl transferase 1A deficiency
- Cerebrotendinous xanthomatosis
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- Cholesteryl ester storage disease
- Chronic visceral acid sphingomyelinase deficiency
- Citrullinemia type II
- Congenital sialidosis type 2
- Congenital sucrase-isomaltase deficiency with starch and lactose intolerance
- Glycogen storage disease IV
- Glycogen storage disease due to glycogen branching enzyme deficiency, progressive hepatic form
- Infantile neurovisceral acid sphingomyelinase deficiency
- Juvenile sialidosis type 2
- McCune-Albright syndrome
- Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency
- Mucopolysaccharidosis type 7
- NON RARE IN EUROPE: Heterozygous familial hypercholesterolemia
- Neonatal intrahepatic cholestasis due to citrin deficiency
- 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
- Primary Fanconi renotubular syndrome
- Sialidosis type 1

- Smith-Lemli-Opitz syndrome
- Transaldolase deficiency
- Wilson disease
- Wolman disease

Related Laboratories

- Centre de Génétique Médicale UCL

Related Analytes

- ATPase copper transporting beta
- carnitine palmitoyltransferase 1A
- cytochrome P450 family 27 subfamily A member 1
- deoxyguanosine kinase
- 7-dehydrocholesterol reductase
- enoyl-CoA hydratase and 3-hydroxyacyl CoA dehydrogenase
- 1,4-alpha-glucan branching enzyme 1
- GNAS complex locus
- glucuronidase beta
- lipase A, lysosomal acid type
- mitochondrial inner membrane protein MPV17
- neuraminidase 1
- NPC intracellular cholesterol transporter 1
- NPC intracellular cholesterol transporter 2
- DNA polymerase gamma, catalytic subunit
- sucrase-isomaltase
- solute carrier family 25 member 13
- sphingomyelin phosphodiesterase 1
- transaldolase 1

- tRNA mitochondrial 2-thiouridylase

Related Gene Panels

- Metabolic diseases with hepatic disorders (20 genes) - UCL
