GENETIC TEST:
Bile Acid Synthesis Congenital Defect (gene panel)

FULL NAME:	Bile Acid Synthesis Congenital Defect (gene panel)
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:	565471-565482
ACCREDITATION (ISO 15189):	2022-12-22 / 2027-12-21
TURNAROUND TIME (MAXIMUM):	3 months

CREATED:	06 Nov 2019 - 09:06
CHANGED:	24 Jan 2023 - 14:43

RELATED CONTENT

Related Diseases

- Cerebrotendinous xanthomatosis
- Congenital bile acid synthesis defect type 1
- Congenital bile acid synthesis defect type 2
- Congenital bile acid synthesis defect type 3
- Congenital bile acid synthesis defect type 4

Related Laboratories

- Centre de Génétique Médicale UCL

Related Analytes

- aldo-keto reductase family 1 member D1
- alpha-methylacyl-CoA racemase
- cytochrome P450 family 27 subfamily A member 1
- cytochrome P450 family 7 subfamily B member 1
- hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 7

Related Gene Panels

- Bile Acid Synthesis Congenital Defect (5 genes) - UCL

Source URL: http://gentest.healthdata.be/genetic_test/917