GENETIC TEST:
Cholestasis (gene panel)

FULL NAME:	Cholestasis (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Post-natal Diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Chorionic villi, Amniotic fluid
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 - 10:46
CHANGED:	24 Jan 2023 - 15:01

RELATED CONTENT

Related Diseases

- Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins
- Alagille syndrome due to a JAG1 point mutation
- Alagille syndrome due to a NOTCH2 point mutation
- Arthrogryposis, renal dysfunction, and cholestasis
- Arthrogryposis-renal dysfunction-cholestasis syndrome
- Autosomal recessive polycystic kidney disease
- Bardet-Biedl syndrome
- Benign recurrent intrahepatic cholestasis type 2
- Caroli disease
- Cerebrotendinous xanthomatosis
- Chronic visceral acid sphingomyelinase deficiency
- Citrullinemia type II
- 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
- Crigler-Najjar syndrome type 1
- Crigler-Najjar syndrome type 2
- Familial hypercholanemia
- GRACILE syndrome
- HNF1B-related autosomal dominant tubulointerstitial kidney disease
- Infantile nephronophthisis
- Infantile neurovisceral acid sphingomyelinase deficiency
- Intrahepatic cholestasis of pregnancy
- Isolated complex III deficiency
- Isolated neonatal sclerosing cholangitis

- Joubert syndrome
- Joubert syndrome with hepatic defect
- Juvenile nephronophthisis
- Late-onset nephronophthisis
- Meckel syndrome
- Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria
- Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency
- Navajo neurohepatopathy
- Neonatal ichthyosis-sclerosing cholangitis syndrome
- Neonatal intrahepatic cholestasis due to citrin deficiency
- Niemann-Pick disease type C
- 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
- Progressive familial intrahepatic cholestasis type 1
- Progressive familial intrahepatic cholestasis type 2
- Progressive familial intrahepatic cholestasis type 3
- Progressive familial intrahepatic cholestasis type 4
- Progressive familial intrahepatic cholestasis type 5
- Renal tubulopathy-encephalopathy-liver failure syndrome
- Renal-hepatic-pancreatic dysplasia
- Senior-Loken syndrome
- Transaldolase deficiency
- Transient familial neonatal hyperbilirubinemia

Related Laboratories

- Centre de Génétique Médicale UCL

Related Analytes

- ATP binding cassette subfamily B member 11
- ATP binding cassette subfamily B member 4
- aldo-keto reductase family 1 member D1
- alpha-methylacyl-CoA racemase
- ATPase phospholipid transporting 8B1
- bile acid-CoA:amino acid N-acyltransferase
- BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone
- coiled-coil and C2 domain containing 2A
- claudin 1
- cytochrome P450 family 27 subfamily A member 1
- cytochrome P450 family 7 subfamily B member 1
- doublecortin domain containing 2
- deoxyguanosine kinase
- HNF1 homeobox B
- hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 7
- inversin
- jagged canonical Notch ligand 1
- MKS transition zone complex subunit 1
- mitochondrial inner membrane protein MPV17
- notch receptor 2
- NPC intracellular cholesterol transporter 1
- NPC intracellular cholesterol transporter 2
- nephrocystin 1
- nephrocystin 3
- nephrocystin 4
- nuclear receptor subfamily 1 group H member 4
- PKHD1 ciliary IPT domain containing fibrocystin/polyductin
- patatin like phospholipase domain containing 3
- Patatin like phospholipase domain containing 3
- DNA polymerase gamma, catalytic subunit

- solute carrier family 25 member 13
- sphingomyelin phosphodiesterase 1
- transaldolase 1
- tight junction protein 2
- Transmembrane 6 superfamily member 2
- Transmembrane channel like 4
- transmembrane protein 216
- tRNA mitochondrial 2-thiouridylase
- UDP glucuronosyltransferase family 1 member A1
- VPS33B interacting protein, apical-basolateral polarity regulator, spe-39 homolog
- VPS33B late endosome and lysosome associated

Related Gene Panels

- Cholestasis (40 genes) - UCL
