DISEASE:
Transient familial neonatal hyperbilirubinemia

NAME:	Transient familial neonatal hyperbilirubinemia
DESCRIPTION:	A rare genetic hepatic disease characterized by very high serum bilirubin levels in a newborn, clinically presenting as jaundice during the first few days of life. The condition is usually self-resolving, although in some cases it can lead to kernicterus with corresponding symptoms (including lethargy, high-pitched crying, hypotonia, missing reflexes, vomiting, or seizures, among others), which may result in chronic disability and even death.
ORPHACODE:	2312
SYNONYMS:	Lucey-Driscoll syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	UGT1A1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Gilbert disease - UGT1A1*28,*36,*37 {A(TA)nTAA} + *6 genotyping - Pharmacogenetics
- Gilbert syndrome / Irinotecan sensitivity (homozygous A(TA)7TAA allele)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- UDP glucuronosyltransferase family 1 member A1

Related Gene Panels

- Cholestasis (40 genes) - UCL