DISEASE:
Hemoglobinopathy Toms River

NAME:	Hemoglobinopathy Toms River
DESCRIPTION:	Hemoglobinopathy Toms River is a rare, genetic hemoglobinopathy disorder, due to a defect in the gamma subunit of the fetal hemoglobin, characterized by neonatal cyanosis, low hemoglobin oxygen saturation levels without arterial hypoxemia, moderate anemia and reticulocytosis, not associated with heart or lung disease. Symptoms progressively subside within the first months of life.
ORPHACODE:	280615
SYNONYMS:	Transient neonatal cyanosis and anemia due to Toms River Hemoglobin
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	HBG2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Beta-globin hemoglobinopathies, phenotype modifiers (hot spot mutations - rs7482144 (Xmn1) at promoter 158 bp 5? upstream of HBG2 / 32C-T in the 5' UTR of the HBS1L)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB

Related Analytes

- hemoglobin subunit gamma 2

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