DISEASE:
Hemoglobin Lepore-beta-thalassemia syndrome

NAME:	Hemoglobin Lepore-beta-thalassemia syndrome
DESCRIPTION:	A rare beta-thalassemia associated with another hemoglobin anomaly characterized by the presence of the hemoglobin Lepore variant in association with beta-thalassemia. Clinical presentation is highly variable, depending on the type of beta-thalassemia, and ranges from severe hypochromic microcytic anemia and complete transfusion dependency to moderate, compensated anemia without a need for regular blood transfusions.
ORPHACODE:	330032
SYNONYMS:	HbLepore-beta-thalassemia syndrome Lepore-beta-thalassemia syndrome
XREF(S):	Orphanet
ANALYTE(S):	HBB HBD
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Beta-globin hemoglobinopathies

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB

Related Analytes

- hemoglobin subunit beta
- hemoglobin subunit delta