DISEASE:
Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome

NAME:	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome
DESCRIPTION:	A rare, genetic, hemoglobinopathy characterized by generally mild clinical phenotype, high fetal hemoglobin levels and mild microcytosis and hypochromia. In some cases, acute sickle cell disease manifestations were reported, namely acute chest syndrome and acute pain crisis. The genotype is characterized by the combination of an HbS and HbF allele; symptoms depend on the degree of HbF:HbS expressivity with patients with more than 35% pancellular HbF expression being asymptomatic. Symptomatic patients have heterocellular expression of HbF.
ORPHACODE:	251380
SYNONYMS:	HPFH-sickle cell disease syndrome
XREF(S):	Orphanet OMIM ICD-10 OMIM OMIM OMIM OMIM

ANALYTE(S):	HBB BCL11A HBG1 HBG2 KLF1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/939

RELATED CONTENT

Related Genetic Tests

- Beta-globin hemoglobinopathies
- Beta-globin hemoglobinopathies
- 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
- Centre de Génétique Médicale UCL

Related Analytes

- BCL11 transcription factor A
- hemoglobin subunit beta
- hemoglobin subunit gamma 1
- hemoglobin subunit gamma 2
- KLF transcription factor 1