DISEASE:
Hermansky-Pudlak syndrome due to BLOC-3 deficiency

NAME:	Hermansky-Pudlak syndrome due to BLOC-3 deficiency
DESCRIPTION:	Hermansky-Pudlak syndrome with pulmonary fibrosis as a complication includes two types (HPS-1 and HPS-4) of Hermansky-Pudlak syndrome (HPS; see this term), a multi-system disorder characterized by oculocutaneous albinism, bleeding diathesis and, in some cases, pulmonary fibrosis or granulomatous colitis.
ORPHACODE:	231500
SYNONYMS:	HPS with pulmonary fibrosis Hermansky-Pudlak syndrome with pulmonary fibrosis
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	HPS1 HPS4
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Pulmonary Fibrosis (gene panel) + rs35705950 of MUC5B gene
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- HPS1 biogenesis of lysosomal organelles complex 3 subunit 1
- HPS4 biogenesis of lysosomal organelles complex 3 subunit 2

Related Gene Panels

- Pulmonary Fibrosis (21 genes) + rs35705950 (MUC5B gene) - KUL
- Trombosis - Hemostasis (107 genes) - KUL