

DISEASE:**Hermansky-Pudlak syndrome due to BLOC-2 deficiency**

NAME:	Hermansky-Pudlak syndrome due to BLOC-2 deficiency
DESCRIPTION:	Hermansky-Pudlak syndrome without pulmonary fibrosis as a complication includes three relatively mild types (HPS-3, HPS-5 and HPS-6) of Hermansky-Pudlak syndrome (HPS; see this term), a multi-system disorder characterized by ocular or oculocutaneous albinism, bleeding diathesis and, in some cases, granulomatous colitis.
ORPHACODE:	231512
SYNONYMS:	HPS without pulmonary fibrosis Hermansky-Pudlak syndrome without pulmonary fibrosis
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u> <u>OMIM</u> <u>OMIM</u>
ANALYTE(S):	<u>HPS3</u> <u>HPS5</u> <u>HPS6</u>
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