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

NAME:	Hermansky-Pudlak syndrome due to AP-3 deficiency
DESCRIPTION:	Hermansky-Pudlak syndrome type 2 (HPS-2) is a type of Hermansky-Pudlak syndrome (HPS; see this term), a multi-system disorder characterized by oculocutaneous albinism, bleeding diathesis and neutropenia.
ORPHACODE:	183678
SYNONYMS:	Hermansky-Pudlak syndrome with neutropenia
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	AP3B1
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