

Opis choroby *

Definicja

Hermansky-Pudlak syndrome (HPS) is a multi-system disorder characterized by oculocutaneous albinism, bleeding diathesis and, in some cases, neutropenia, pulmonary fibrosis, or granulomatous colitis. HPS comprises eight known disorders (HPS-1 to HPS-8), the majority of which present with the same clinical phenotype to varying degrees of severity.

Dane

Klasyfikacja

Choroba

Synonimy

HPS

HPS

Kod ORPHA

79430

Kod OMIM

614171

Kod ICD10

E70.3

Kod ICD11

EC23.20

*Źródło

orphanet