Hermansky-Pudlak syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hermansky-Pudlak syndrome. ORPHA:79430

Hermansky-Pudlak syndrome (HSP) 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.