H syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. H syndrome. ORPHA:168569

H syndrome is a systemic inherited histiocytosis, with characteristic cutaneous findings accompanying systemic manifestations. H syndrome refers to the major clinical findings of hyperpigmentation, hypertrichosis, hepatosplenomegaly, heart anomalies, hearing loss, hypogonadism, low height, and occasionally, hyperglycemia/diabetes mellitus. Due to overlapping clinical features, H syndrome is now considered to include pigmented hypertrichosis with insulin dependent diabetes mellitus syndrome (PHID), Faisalabad histiocytosis (FHC) and familial sinus histiocytosis with massive lymphadenopathy (FSHML).