DISEASE:
Hereditary pulmonary alveolar proteinosis

NAME:	Hereditary pulmonary alveolar proteinosis
DESCRIPTION:	A rare, genetic, interstitial lung disease due to mutations in the CSF2R (colony-stimulating factor 2 receptor) alpha or beta subunits and characterized by alveolar accumulation of pulmonary surfactant, presenting a highly variable clinical presentation, ranging from asymptomatic to severe respiratory failure. Characteristic lung biopsy findings include periodic acid-Schiff-positive, granular eosinophilic material, enlarged foamy alveolar macrophages, and well-preserved alveolar walls. The Granulocyte-macrophage colony-stimulating factor (GM-CSF) receptor function is impaired but GM-CSF receptor autoantibodies are absent.
ORPHACODE:	264675
SYNONYMS:	Congenital PAP Congenital pulmonary alveolar proteinosis
XREF(S):	Orphanet OMIM ICD-10 OMIM
ANALYTE(S):	CSF2RB CSF2RA
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