

[Open Peer Review on Qeios](#)

Severe early-onset pulmonary alveolar proteinosis due to MARS deficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Severe early-onset pulmonary alveolar proteinosis due to MARS deficiency. ORPHA:440427*

A rare, genetic interstitial lung disease characterized by accumulation of lipoproteins in the pulmonary alveoli leading to restrictive lung disease and respiratory failure. Patients present with dyspnea, tachypnea, cough, failure to thrive, and digital clubbing. Liver disease have been described in some cases including hepatomegaly, steatosis, fibrosis or cirrhosis.