

Opis choroby *

Definicja

A rare genetic interstitial lung disease characterized by progressive, life-threatening, refractory respiratory distress in full-term neonates associated with surfactant protein B deficiency. In most cases, the disease is fatal within the first months of life. Lung biopsy reveals changes characteristic of pulmonary alveolar proteinosis with interstitial fibrosis and inflammation, as well as accumulation of lipid-rich, eosinophilic, proteinaceous, granular material consisting of desquamated type II pneumocytes and foamy macrophages within the alveolar air spaces.

Dane

Klasyfikacja

Choroba

Synonimy

Neonatal acute respiratory distress due to surfactant protein B deficiency
Ostra niewydolność oddechowa noworodków spowodowana niedoborem białka B surfaktantu

Kod ORPHA

217563

Kod OMIM

265120

Kod ICD10

P28.0

Kod ICD11

CB04.2

*Źródło

orphanet