Opis choroby *

Definicja

A rare genetic interstitial lung disease characterized by diffuse lung disease of variable phenotype ranging from severe respiratory insufficiency in infancy to asymptomatic adults, due to surfactant protein C deficiency. Typical presentation in infancy includes dyspnea, cough, wheezing, and gradual cyanosis, with or without failure to thrive. Radiological findings include diffuse ground-glass opacities in neonates, later interstitial thickening associated with lung hyperinflation, intraparenchymal/subpleural cysts, honeycombing, subpleural nodules, or bronchiectasis. Infiltrates and air leaks are frequent complications.

Dane

Klasyfikacja
Choroba

Synonimy
Interstitial lung disease due to surfactant protein C deficiency
Śródmiąższowa choroba płuc spowodowana niedoborem białka C surfaktantu

Kod ORPHA
440392

Kod OMIM
610913

Kod ICD10
J84.8

Kod ICD11
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*Źródło

orphanet