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 Synonimy

Choroba Interstitial lung disease due to surfactant protein

C deficiency

Śródmiąższowa choroba płuc spowodowana

niedoborem białka C surfaktantu

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 440392
 610913
 I84.8

Kod ICD11

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*Źródło

orphanet