

## Opis choroby \*

### Definicja

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.

### Dane

Klasyfikacja	Synonimy	
Choroba	Congenital PAP	
	Wrodzona PAP	
	Congenital pulmonary alveolar proteinosis	
<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
264675	300770	J84.0

### Kod ICD11

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### \*Źródło

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