Opis choroby *

Definicja

A rare genetic respiratory disease characterized by widespread intra-alveolar accumulation of minute calcium phosphate microliths, leading to pulmonary fibrosis, pulmonary hypertension, and chronic respiratory failure. Age of onset is highly variable, and most patients are asymptomatic for years or decades, before signs and symptoms like dyspnea on exertion, dry cough, chest pain, hemoptysis, or finger clubbing develop. The disease takes a long-term progressive course. Routine chest radiographs typically show a fine, ''sandstorm-like'' micronodular pattern that is more pronounced in the bases than in the apices.

Dane

Klasyfikacja
Choroba

Kod ORPHA
60025

Kod OMIM
265100

Kod ICD10
J84.0

Kod ICD11
CB06

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*Źródło

orphanet