

Opis choroby *

Definicja

Interstitial lung disease due to ABCA3 deficiency is a rare genetic respiratory disease characterized by a variable clinical outcome ranging from a fatal respiratory distress syndrome in the neonatal period to chronic interstitial lung disease developing in infancy or childhood with chronic cough, rapid breathing, shortness of breath and recurrent pulmonary infections. Clinical manifestations of respiratory failure include grunting, intercostal retractions, nasal flaring, cyanosis, and progressive dyspnea.

Dane

Klasyfikacja

Choroba

Synonimy

Interstitial lung disease due to ATP-binding cassette subfamily A member 3 deficiency
Śródmiąższowa choroba płuc spowodowana niedoborem członka 3 podrodziny A kasety wiążącej ATP

Kod ORPHA

440402

Kod OMIM

610921

Kod ICD10

J84.8

Kod ICD11

-

*Źródło

orphanet