Opis choroby *

Definicja

A rare genetic respiratory disease characterized by infantile onset of pulmonary alveolar proteinosis with hypogammaglobulinemia. Patients have normal respiratory function at birth, but subsequently develop recurrent, mainly viral, infections and progressive respiratory failure, often leading to death in infancy or early childhood. Additional reported features include leukocytosis and splenomegaly.

Dane

Klasyfikacja
Choroba

Synonimy
OAS1 deficiency
Proteinoza pęcherzyków płucnych i hipogammaglobulinemia o początku w niemowlęctwie związana z OAS1
OAS1-related infantile-onset pulmonary alveolar proteinosis-hypogammaglobulinemia

Kod ORPHA
572428

Kod OMIM
-

Kod ICD10
J84.0

Kod ICD11
-

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*Źródło

orphanet