

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

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Kod ICD10

J84.0

Kod ICD11

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*Źródło

orphanet