

Opis choroby *

Definicja

A rare, syndromic, genetic respiratory disease characterized by cataracts, otitis media, intestinal malabsorption, chronic respiratory infections, and failure to thrive. Recurrent pneumonia and progressive azotemia, leading to end-stage renal disease and early death, are additionally observed. There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

3167

Kod OMIM

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Kod ICD10

J98.8

Kod ICD11

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*Źródło

orphanet