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