Opis choroby *

Definicja

A life-threatening multiorgan disorder which develops in the first months of life, presenting with respiratory distress and proteinuria in the nephrotic range, and leading to severe interstitial lung disease and renal failure. Some patients additionally display cutaneous alterations, ranging from blistering and skin erosions to an epidermolysis bullosa-like phenotype, with toe nail dystrophy and sparse hair.

Dane

Klasyfikacja
Choroba

Synonimy
ILNEB syndrome
JEB with interstitial lung disease and nephrotic syndrome
Junctional epidermolysis bullosa with interstitial lung disease and nephrotic syndrome
ILNEB syndrome
JEB with interstitial lung disease and nephrotic syndrome
Junctional epidermolysis bullosa with interstitial lung disease and nephrotic syndrome

Kod ORPHA
306504

Kod OMIM
614748

Kod ICD10
J84.8

Kod ICD11
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*Źródło

orphanet