

Wrodzony ubytek skóry - limfangiectazja jelitowa

Kod Orpha: 1116 Kod OMIM: 207731

Opis choroby *

Definicja

An extremely rare association syndrome, described in only two brothers to date (one of which died at 2 months of age), characterized by aplasia cutis congenita of the vertex and generalized edema (as well as hypoproteinemia and lymphopenia) due to intestinal lymphangiectasia. There have been no further descriptions in the literature since 1985.

Dane

Klasyfikacja

Choroba

Synonimy

Bronspiegel-Zelnick syndrome

Autosomalna recesywna aplazja skóry

Zespół Bronspiegela i Zelnicka

Kod ORPHA

1116

Kod OMIM

207731

Kod ICD10

Q84.8

Kod ICD11

LD27.Y

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.