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

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*Źródło

orphanet