Opis choroby *

Definicja

A rare, inherited, epidermolysis bullosa simplex characterized by neonatal onset of generalized or, less frequently, localized acral blistering. Milia are rare but atrophic scarring and dystrophic nails usually occur, along with focal keratoderma (palms and soles). Severe generalized blistering may cause perinatal death or persist during the entire life. Extracutaneous involvement is common, including anemia, growth retardation, oral cavity abnormalities (blisters and erosions, and caries) and constipation.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive generalized EBS
Pęcherzowe oddzielanie się naskórka, postać prosta autosomalna recesywna

Kod ORPHA
89838

Kod OMIM
601001

Kod ICD10
Q81.0

Kod ICD11
EC30

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*Źródło

orphanet