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			
<u>*Źródło</u>			
orphanet			