

Opis choroby *

Definicja

An X-linked syndromic multi-systemic ectodermal dysplasia presenting neonatally in females with a bullous rash along Blaschko's lines (BL) followed by verrucous plaques and hyperpigmented swirling patterns. It is further characterized by teeth abnormalities, alopecia, nail dystrophy and can affect the retinal and the central nervous system (CNS) microvasculature. It may have other aspects of ectodermal dysplasia such as sweat gland abnormalities. Germline pathogenic variants in males result in embryonic lethality.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Bloch-Siemens syndrome Zespół Blocha i Simonsa Zespół Blocha i Sulzbergera Bloch-Sulzberger syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
464	308300	Q82.3

Kod ICD11
LD27.00

*Źródło

orphanet