

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Bloch-Siemens syndrome
	Zespół Blocha i Simonsa
	Zespół Blocha i Sulzbergera
	Bloch-Sulzberger syndrome

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
464	308300	Q82.3

**Kod ICD11**  
LD27.00

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### \*Źródło

orphanet