

## Opis choroby \*

### Definicja

A very rare syndrome characterized by a combination of blepharochalasis, double lip, and non-toxic thyroid enlargement (seen in 10-50% of cases), although the occurrence of all three signs at presentation is uncommon. Hypertrophy of the mucosal zone of the lip with persistence of the horizontal sulcus between cutaneous and mucosal zones gives an appearance of double lip, with the upper lip being frequently involved. Blepharochalasis, or episodic edema of eyelid, appears around puberty, is present in 80% of cases, is usually bilateral, and can rarely lead to vision impairment and other ocular complications. Most cases are sporadic, but familial cases (with a possible autosomal dominant inheritance) have also been reported.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Blepharochalasis-double lip syndrome  
Zespół zwiotczenia powiek i podwójnej wargi

#### Kod ORPHA

1253

#### Kod OMIM

109900

#### Kod ICD10

Q87.0

#### Kod ICD11

LD27.Y

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#### \*Źródło

orphanet