

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

*Źródło

orphanet