Opis choroby *

Definicja

A rare, genetic, superficial corneal dystrophy disease characterized by white, elevated, epithelial plaques located on the bulbar conjunctiva (sometimes with encroachment of the cornea) and oral mucosa (in any part of the oral cavity), associated with dilated, hyperemic, conjunctival blood vessels, observed mainly in Haliwa-Saponi Native American descendents. Patients may be asymptomatic or present with ocular itching, superficial corneal scarring, excessive lacrimation, photophobia and visual loss due to corneal opacity. Histologically, both ocular and oral lesions display acanthosis with hyperkeratosis and prominent dyskeratosis.

Dane

Klasyfikacja
Choroba

Synonimy
HBID
Dziedziczna łagodna dyzkeratoza wewnątrznabłonkowa rogówki
HBID
Hereditary benign corneal intraepithelial dyskeratosis

Kod ORPHA
352657

Kod OMIM
127600

Kod ICD10
Q82.8

Kod ICD11
DA02.0

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*Źródło

orphanet