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 Synonimy Choroba HBID

Dziedziczna łagodna dyzkeratoza wewnątrznabłonkowa rogówki

HBID

Hereditary benign corneal intraepithelial

dyskeratosis

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 352657
 127600
 Q82.8

Kod ICD11 DA02.0

*Źródło

orphanet