

Opis choroby *

Definicja

A rare genetic disease characterized by sensorineural hearing loss, abnormalities in the secondary dentition (such as enamel hypoplasia, taurodontism, or dental overcrowding), and nail abnormalities (including leukonychia and presence of transverse ridges). Association with macular dystrophy has also been reported.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Hearing loss-enamel hypoplasia-nail defects syndrome Zespół Heimlera Heimler syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
3220	616617	Q82.4

Kod ICD11
LD27.0Y

*Źródło

orphanet