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
Zespół wad wrodzonych

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

Kod ORPHA
3220

Kod OMIM
616617

Kod ICD10
Q82.4

Kod ICD11
LD27.0Y

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*Źródło

orphanet