Opis choroby *

Definicja

A rare malformative syndrome with dentinogenesis imperfecta, characterized by dentin dysplasia with opalescent discoloration and severe attrition of primary and permanent teeth, and delayed eruption, bulbous crowns, long and tapered roots, and progressive root canal obliteration of the permanent dentition, associated with proportionate short stature, sensorineural hearing loss, mild intellectual disability, and dysmorphic facial features. The latter include a prominent nose with high nasal bridge and short philtrum. Osteoporosis, mild platyspondyly, and cone-shaped epiphyses have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dentinogenesis imperfecta-short stature-deafness-intellectual disability syndrome
Dentinogenesis imperfecta-short stature-deafness-intellectual disability syndrome

Kod ORPHA
71267

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
LD2H.Y

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*Źródło

orphanet