

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Dentinogenesis imperfecta-short stature-deafness-intellectual disability syndrome Dentinogenesis imperfecta-short stature-deafness-intellectual disability syndrome

<b>Kod ORPHA</b> 71267	<b>Kod OMIM</b> -	<b>Kod ICD10</b> Q87.8
<b>Kod ICD11</b> LD2H.Y		

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### \*Źródło

orphanet