Opis choroby *

Definicja

A extremely rare, genetic malformation syndrome characterized by hypoplastic amelogenesis imperfecta (hypoplastic dental enamel) and nephrocalcinosis (precipitation of calcium salts in renal tissue). Oral manifestations include yellow and misshaped teeth, delayed tooth eruption, and intrapulpal calcifications. Nephrocalcinosis is often asymptomatic but can progress during late childhood or early adulthood to impaired renal function, recurrent urinary infections, renal tubular acidosis, and rarely to end-stage renal failure.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Amelogenesis imperfecta-nephrocalcinosis syndrome

Kod ORPHA
1031

Kod OMIM
204690

Kod ICD10
K00.5

Kod ICD11
LA30.6

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*Źródło

orphanet