

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

Synonimy

Zespół wad wrodzonych Amelogenesis imperfecta-nephrocalcinosis syndrome

Kod ORPHA

1031

Kod OMIM

204690

Kod ICD10

K00.5

Kod ICD11

LA30.6

*Źródło

orphanet