

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

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|-----------------------|---------------------------------------------------|
| Klasyfikacja | Synonimy |
| Zespół wad wrodzonych | Amelogenesis imperfecta-nephrocalcinosis syndrome |

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|------------------|-----------------|------------------|
| Kod ORPHA | Kod OMIM | Kod ICD10 |
| 1031 | 204690 | K00.5 |

Kod ICD11
LA30.6

*Źródło

orphanet