## Opis choroby *

Definicja

A rare hypomyelinating leukodystrophy disorder characterized by the association of dental abnormalities (delayed dentition, abnormal order of dentition, hypodontia), hypogonadotropic hypogonadism, and hypomyelinating leukodystrophy manifesting with neurodevelopmental delay or regression and/or progressive cerebellar symptoms.

Dane

**Klasyfikacja**
Choroba

Synonimy
POLR-related leukodystrophy
Leukodystrofia związna z Pol III

**Kod ORPHA**
289494

**Kod OMIM**
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**Kod ICD10**
G37.8

**Kod ICD11**
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*Źródło

orphanet