Opis choroby *

Definicja

A rare, primary bone dysplasia characterized by prenatal and postnatal growth retardation, short stature, cortical thickening and medullary stenosis of the long bones, absent diploic space in the skull bones, hypocalcemia due to the hypoparathyroidism, small hands and feet, delayed mental and motor development, intellectual disability, dental anomalies, and dysmorphic features, including prominent forehead, small deep-set eyes, beaked nose, and micrognathia.

Dane

Klasyfikacja
Podtyp etiologiczny

Kod ORPHA
93324

Kod OMIM
244460

Kod ICD10
Q87.1

Kod ICD11
LD24.D

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*Źródło

orphanet