Opis choroby *

Definicja

A rare primary bone dysplasia characterized by global developmental delay, hypotonia, ossification anomalies of the cranial vault, abnormalities of the long bones due to defective remodeling, thoracic deformity, and progressive osteopenia. Dysmorphic craniofacial features include microcephaly, hypertelorism, narrow mouth, cleft palate, and micrognathia.

Dane

Klasyfikacja
Choroba

Kod ORPHA
73230

Kod OMIM
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Kod ICD10
Q79.8

Kod ICD11
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*Źródło

orphanet