

Opis choroby *

Definicja

A primary bone dysplasia with micromelia characterized by rhizomelia, exaggerated lumbar lordosis, brachydactyly, and macrocephaly with frontal bossing and midface hypoplasia.

Dane

Klasyfikacja

Choroba

Kod ORPHA

15

Kod OMIM

100800

Kod ICD10

Q77.4

Kod ICD11

LD24.00

*Źródło

orphanet