Opis choroby *

Definicja

A rare, primary bone dysplasia characterized by proportional short stature, early cessation of bone growth, accelerated skeletal maturation, variable presence of early-onset osteoarthritis and osteochondritis dissecans, and normal endocrine evaluation. The variable dysmorphic features include mild to relative macrocephaly, frontal bossing, midfacial hypoplasia, flat nasal bridge, brachydactyly, broad thumbs, and lordosis.

Dane

Klasyfikacja
Choroba

Kod ORPHA
435804

Kod OMIM
165800

Kod ICD10
M89.8

Kod ICD11
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*Źródło

orphanet