

## 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