

## Opis choroby \*

### Definicja

A rare, lethal type of achondrogenesis, and part of the spectrum of type 2 collagen-related bone disorders, characterized by severe micromelia, short neck with large head, small thorax, protuberant abdomen, underdeveloped lungs, distinctive facial features such as a prominent forehead, a small chin, a cleft palate (in some) and distinctive histological features of the cartilage.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Achondrogenesis, Langer-Saldino type  
Achondrogeneza typu Langer i Saldino

#### Kod ORPHA

93296

#### Kod OMIM

200610

#### Kod ICD10

Q77.0

#### Kod ICD11

LD24.50

---

#### \*Źródło

orphanet