

## Opis choroby \*

### Definicja

A rare, lethal type of achondrogenesis characterized by severe micromelia with very short fingers and toes, a flat face, a short neck, thickened soft tissue around the neck, hypoplasia of the thorax, protuberant abdomen, a hydropic fetal appearance and distinctive histological features of the cartilage.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Achondrogenesis, Parenti-Fraccaro type  
Achondrogeneza typu Parenti i Fraccaro

#### Kod ORPHA

93298

#### Kod OMIM

600972

#### Kod ICD10

Q77.0

#### Kod ICD11

LD24.50

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#### \*Źródło

orphanet