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

---

*Źródło

orphanet