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 Langera i Saldino

Kod ORPHA
93296

Kod OMIM
200610

Kod ICD10
Q77.0

Kod ICD11
LD24.50

---

*Źródło

orphanet