

Achondrogenesa typu 2

Kod Orpha: 93296 Kod OMIM: 200610

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
Achondrogenesa typu Langera i Saldino

Kod ORPHA

93296

Kod OMIM

200610

Kod ICD10

Q77.0

Kod ICD11

LD24.50

*[Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.