

Opis choroby *

Definicja

A rare group of lethal skeletal dysplasias characterized by an endochondral ossification deficiency that leads to dwarfism with extreme micromelia, a small thorax, a prominent abdomen, anasarca and polyhydramnios. There are three types of achondrogenesis that exist and that differ clinically, radiologically, histologically and genetically: achondrogenesis type 1a, type 1b and type 2.

Dane

Klasyfikacja

Choroba

Kod ORPHA

932

Kod OMIM

600972

Kod ICD10

Q77.0

Kod ICD11

LD24.50

*Źródło

orphanet