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: achondrogensis type 1a, type 1b and type 2.

Dane

Klasyfikacja
Choroba

Kod ORPHA
932

Kod OMIM
600972

Kod ICD10
Q77.0

Kod ICD11
LD24.50

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*Źródło

orphanet