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

<u>*Źródło</u>

orphanet