

Opis choroby *

Definicja

A rare, genetic, primary bone dysplasia with decreased bone density characterized by fetal lethality, severe hypomineralization of the entire skeleton, barrel shaped thorax with short ribs, multiple intrauterine fractures of ribs and long bones, ascites, pleural effusion, and ventriculomegaly. Variable congenital developmental anomalies affecting the brain, lungs, and kidneys have also been associated.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Complex lethal osteochondrodysplasia,
Symoens-Barnes-Gistelinck type
Złożona śmiertelna osteochondrodysplazja typu
Symoensa, Barnesa i Gistelincka

Kod ORPHA

457378

Kod OMIM

616897

Kod ICD10

Q78.8

Kod ICD11

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*Źródło

orphanet