Śmiertelna recesywna chondrodysplazja
Kod Orpha: 1423 Kod OMIM:

Opis choroby *

Definicja
Lethal recessive chondrodysplasia is an extremely rare lethal form of chondrodysplasia characterized by severe micromelic dwarfism, short and incurved limbs with normal hands and feet, facial dysmorphism (disproportionately large skull, frontal prominence, slightly flattened nasal bridge and short neck), muscular hypotonia, hyperlaxity of the extremities, and a narrow thorax. Most patients die of respiratory distress during the first hours or weeks of life. There have been no further descriptions in the literature since 1988.
Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Maroteaux-Stanescu-Cousin syndrome
Zespół Maroteaux, Stanescu i Cousina

Kod ORPHA
1423

Kod OMIM
-

Kod ICD10
Q78.8

Kod ICD11
FB82.Y

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.