Fibrochondrogeneza
Kod Orpha: 2021 Kod OMIM: 614524

Opis choroby *

Definicja
Fibrochondrogenesis is a rare, neonatally lethal, rhizomelic chondrodysplasia. Eleven cases have been reported. The face is distinctive and characterized by protuberant eyes, flat midface, flat small nose with anteverted nares and a small mouth with long upper lip. Cleft palate, micrognathia and bifid tongue can occur. The limbs show marked shortness of all segments with relatively normal hands and feet. No internal anomalies other than omphalocele have been reported. Transmission is probably autosomal recessive. Recurrence in a consanguineous family (affecting both sexes) and concordance of affected male twins have been reported.
Dane

Klasyfikacja
Choroba

Kod ORPHA
2021

Kod OMIM
614524

Kod ICD10
Q77.7

Kod ICD11
LD24.3

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.