Diaphanospondylodysostosis
Kod Orpha: 66637 Kod OMIM: 608022

Opis choroby *

Definicja
Diaphanospondylodysostosis is characterized by absent ossification of the vertebral bodies and sacrum associated with variable anomalies. It has been described in less than ten patients from different families. Manifestations include a short neck, a short wide thorax, a reduced number of ribs, a narrow pelvis, and inconstant anomalies such as myelomeningocele, cystic kidneys with nephrogenic rests, and cleft palate.
Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
66637

Kod OMIM
608022

Kod ICD10
Q78.8

Kod ICD11
LD24.5Y

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.