

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

*Źródło

orphanet