

# 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.