

# Dysplazja pseudodiastroficzna

Kod Orpha: 85174 Kod OMIM: 264180

## Opis choroby \*

### Definicja

Pseudodiastrophic dysplasia is characterized by rhizomelic shortening of the limbs and severe clubfoot deformity, in association with elbow and proximal interphalangeal joint dislocations, platyspondyly, and scoliosis. It has been described in about 10 patients. An autosomal recessive inheritance has been suggested. Pseudodiastrophic dysplasia differs from diastrophic dysplasia (see this term) on the basis of clinical, radiographic, and histopathologic findings. Clubfoot can be treated by surgical therapy, and neonatal contractures and scoliosis can be relieved by physical therapy. Several of the reported patients died in the neonatal period or during infancy.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA  
85174

Kod OMIM  
264180

Kod ICD10  
Q78.8

Kod ICD11  
LD24.E

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[\\*Źródło](#)

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## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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