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

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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