Dysplazja wielonasadowa typu 4
Kod Orpha: 93307 Kod OMIM: 226900

Opis choroby *

Definicja
Multiple epiphyseal dysplasia type 4 is a multiple epiphyseal dysplasia with a late-childhood onset, characterized by joint pain involving hips, knees, wrists, and fingers with occasional limitation of joint movements, deformity of hands, feet, and knees (club foot, clinodactyly, brachydactyly), scoliosis and slightly reduced adult height. Radiographs display flat epiphyses with early arthritis of the hip, and double-layered patella. Multiple epiphyseal dysplasia type 4 follows an autosomal recessive mode of transmission. The disease is allelic to diastrophic dwarfism, atelosteogenesis type 2 and achondrogenesis type 1B with whom it forms a clinical continuum.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive multiple epiphyseal dysplasia
Autosomalna recesywna dysplazja wielonasadowa
EDM4
MED4
rMED
EDM4
MED4
Polyepiphyseal dysplasia type 4
rMED

Kod ORPHA
93307

Kod OMIM
226900

Kod ICD10
Q77.3

Kod ICD11
LD24.61

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.