

## 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](#)

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