

## Opis choroby \*

### Definicja

Multiple epiphyseal dysplasia type 1 (MED 1) is a form of multiple epiphyseal dysplasia that is characterized by normal or mild short stature, pain in the hips and/or knees, progressive deformity of extremities and early-onset osteoarthritis. Specific features to MED 1 include a more pronounced involvement of hip joints and gait abnormality and a shorter adult height. MED1 is allelic to pseudoachondroplasia with which it shares clinical and radiological features. The disease follows an autosomal dominant mode of transmission.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

EDM1

Dysplazja wielonasadowa typu 1

EDM1

MED1

MED1

Polyepiphyseal dysplasia type 1

#### Kod ORPHA

93308

#### Kod OMIM

132400

#### Kod ICD10

Q77.3

#### Kod ICD11

LD24.61

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#### [\\*Źródło](#)

orphanet