Opis choroby *

Definicja

Multiple epiphyseal dysplasia type 5 is a multiple epiphyseal dysplasia characterized by an early-onset of pain and stiffness (involving knee and hip), progressive deformity of the extremities and precocious osteoarthritis associated with delayed and irregular ossification of epiphyses. Features specific to multiple epiphyseal dysplasia, type 5 include normal stature and lesser incidence of gait abnormalities. Radiographs reveal epiphyseal and metaphyseal irregularities. Multiple epiphyseal dysplasia type 5 follows an autosomal dominant mode of transmission.

Dane

Klasyfikacja
Choroba

Synonimy
BHMED
EDM5
MED5
BHMED
Bilateral hereditary micro-epiphyseal dysplasia
EDM5
MED5
Polyepiphyseal dysplasia type 5

Kod ORPHA
93311

Kod OMIM
607078

Kod ICD10
Q77.3

Kod ICD11
LD24.61

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*Źródło

orphanet