

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

[*Źródło](#)

orphanet