Opis choroby *

Definicja

A rare group of primary bone dysplasia disorders characterized by the association of epiphyseal anomalies of long bones causing joint pain early in life, recurrent osteochondritis and early arthrosis. This group contains an heterogeneous group of diseases with variable expression. Common reported clinical signs include waddling gait and pain at onset, and moderate short stature. Some forms are mainly limited to the femoral epiphyses, while several other syndromes are characterized by the association of multiple epiphyseal dysplasia with other clinical manifestations such as myopia, deafness and facial dysmorphism. Diagnosis relies on identification of the radiological features.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
EDM
Dysplazja wielonasadowa
EDM
MED
MED
Polyepiphyseal dysplasia

Kod ORPHA
251

Kod OMIM
-

Kod ICD10
Q77.3

Kod ICD11
LD24.61

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*Źródło

orphanet