

Dysplazja wielonasadowa

Kod Orpha: 251 Kod OMIM:

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

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Kod ICD10

Q77.3

Kod ICD11

LD24.61

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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