Opis choroby *

Definicja

A rare primary bone dysplasia characterized by the association of multiple epiphyseal dysplasia, visual impairment (with early-onset progressive myopia, retinal thinning, and cataracts), and conductive hearing loss. Patients are of short stature and present brachydactyly, genu valgus deformity, and joint pain.

Dane

Klasyfikacja
Choroba

Synonimy
Multiple epiphyseal dysplasia-myopia-deafness syndrome
Dysplazja wielonasadowa - miopia - głuchota
Multiple epiphyseal dysplasia-myopia-hearing loss syndrome

Kod ORPHA
166011

Kod OMIM
132450

Kod ICD10
Q77.3

Kod ICD11
LD24.61

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*Źródło

orphanet