

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

*Źródło

orphanet