

Opis choroby *

Definicja

Multiple epiphyseal dysplasia due to collagen 9 anomaly is a rare primary bone dysplasia disorder characterized by normal or mild short stature, early-onset pain and/or stiffness of the joints (mainly affecting knees but also elbows, wrists, ankles and fingers, with relative sparing of the hips) and early degenerative joint disease. Other skeletal anomalies (incl. varus or valgus deformities, osteochondritis dissecans, abnormal carpal shape, free articular bodies) and mild myopathy have also been reported.

Dane

Klasyfikacja

Choroba

Kod ORPHA

166002

Kod OMIM

614135

Kod ICD10

Q77.3

Kod ICD11

LD24.61

*Źródło

orphanet