

Opis choroby *

Definicja

Multiple epiphyseal dysplasia type 1 (MED 1) is a form of multiple epiphyseal dysplasia that is characterized by normal or mild short stature, pain in the hips and/or knees, progressive deformity of extremities and early-onset osteoarthritis. Specific features to MED 1 include a more pronounced involvement of hip joints and gait abnormality and a shorter adult height. MED1 is allelic to pseudoachondroplasia with which it shares clinical and radiological features. The disease follows an autosomal dominant mode of transmission.

Dane

Klasyfikacja

Choroba

Synonimy

EDM1

Dysplazja wielonasadowa typu 1

EDM1

MED1

MED1

Polyepiphyseal dysplasia type 1

Kod ORPHA

93308

Kod OMIM

132400

Kod ICD10

Q77.3

Kod ICD11

LD24.61

[*Źródło](#)

orphanet