Opis choroby *

Definicja

A rare primary bone dysplasia characterized by abnormal bone metabolism with bone pain, deformity, pathological fractures, early conductive hearing loss, and dental abnormalities. Focal bone lesions are typically found in the appendicular skeleton and consist of progressively expanding lytic areas, while generalized disordered bone modeling and altered trabecular pattern are the result of the multifocal, progressive nature of the disease. Age of onset is variable, mode of inheritance is autosomal dominant.

Dane

Klasyfikacja
Choroba

Synonimy
Hereditary expansile polyostotic osteolytic dysplasia
Choroba McCabe'a
Dziedziczna ekspansywna dysplazja osteolityczna
McCabe disease

Kod ORPHA
85195

Kod OMIM
174810

Kod ICD10
M89.5

Kod ICD11
FB86.2

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*Źródło

orphanet