Opis choroby *

Definicja

A moderate form of osteogenesis imperfecta characterized by increased bone fragility and low bone mass that clinically manifests with susceptibility to bone fractures of variable severity, metaphyseal changes at birth, short stature, dislocation of the radial head, mineralized interosseous membranes, hyperplasic callus (occurring more often during periods of more rapid growth), white sclera and absence of dentinogenesis imperfecta.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
OI type 5
OI typu 5

Kod ORPHA
216828

Kod OMIM
610967

Kod ICD10
Q78.0

Kod ICD11
LD24.K0

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*Źródło

orphanet