

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, hyperplastic 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

*Źródło

orphanet