Opis choroby *

Definicja

An extremely rare form of bone dysplasia characterized by the features of osteogenesis imperfecta such as bone fragility associated with multiple fractures, bone deformities (metaphyseal irregularities and bowing of the long bones) and blue sclera, in association with growth failure, craniosynostosis, hydrocephalus, ocular proptosis, and distinctive facial features (e.g. frontal bossing, midface hypoplasia, and micrognathia).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Bone fragility-craniosynostosis-proptosis-hydrocephalus syndrome
Kruchość kości - kraniosynostoza - proptoza - wodogłowie

Kod ORPHA
2050

Kod OMIM
616294

Kod ICD10
Q78.0

Kod ICD11
LD24.KY

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*Źródło

orphanet