

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

[*Źródło](#)

orphanet