

## 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