

## Opis choroby \*

### Definicja

A rare multiple congenital malformations/dysmorphic syndrome characterized by osteogenesis imperfecta with multiple prenatal bone fractures, joint laxity, severe microcephaly, and bilateral cataracts. Additional reported manifestations include dysmorphic facial features (such as blue sclerae, hypertelorism, and low-set ears), lissencephaly, hydrocephalus, and cardiac and genital anomalies. The syndrome is lethal *in utero* or shortly after birth. There have been no further descriptions in the literature since 1978.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

2772

#### Kod OMIM

259410

#### Kod ICD10

Q78.0

#### Kod ICD11

LD24.KY

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### \*Źródło

orphanet