## 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