

Wrodzona łamliwość kości - małogłowie - zaćma

Kod Orpha: 2772 Kod OMIM: 259410

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.