

Opis choroby *

Definicja

X-linked lethal multiple pterygium syndrome is a rare, genetic, developmental defect during embryogenesis characterized by the typical lethal multiple pterygium syndrome presentation (comprising of multiple pterygia, severe arthrogyriposis, cleft palate, cystic hygromata and/or fetal hydrops, skeletal abnormalities and fetal death in the 2nd or 3rd trimester) with an X-linked pattern of inheritance.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

79447

Kod OMIM

312150

Kod ICD10

Q79.8

Kod ICD11

LD26.40

*Źródło

orphanet