

## 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

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### \*Źródło

orphanet