

Opis choroby *

Definicja

A rare developmental defect with connective tissue involvement characterized by multiple joint dislocations, flattened facial appearance, abnormal palmar creases, laryngotracheomalacia, and pulmonary hypoplasia. Additional signs may include a bifid tongue, micrognathia, non-immune hydrops fetalis, and brain dysplasia. The disease is lethal shortly after birth due to respiratory insufficiency.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2371

Kod OMIM

245650

Kod ICD10

Q74.8

Kod ICD11

LD24.E

*Źródło

orphanet