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

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*Źródło

orphanet