

Hipoplazja strzałkowo-łokciowa - wady nerek

Kod Orpha: 2256 Kod OMIM: 228940

Opis choroby *

Definicja

Fibulo-ulnar hypoplasia-renal anomalies syndrome is characterized by fibuloulnar dysostosis with renal anomalies. It has been described in two sibs born to nonconsanguinous parents. The syndrome is lethal at birth (respiratory failure). Clinical manifestations include ear and facial anomalies (including micrognathia), symmetrical shortness of long bones, fibular agenesis and hypoplastic ulna, oligosyndactyly, congenital heart defects, and cystic or hypoplastic kidney. It is transmitted as an autosomal recessive trait.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Saito-Kuba-Tsuruta syndrome
Zespół Saito, Kuba i Tsuruta

Kod ORPHA

2256

Kod OMIM

228940

Kod ICD10

Q87.8

Kod ICD11

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.