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