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 Synonimy

Zespół wad wrodzonych Saito-Kuba-Tsuruta syndrome

Zespół Saito, Kuba i Tsuruta

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 2256
 228940
 Q87.8

Kod ICD11

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*Źródło

orphanet