Opis choroby *

Definicja

Ulbright-Hodes syndrome is characterised by renal dysplasia, growth retardation, phocomelia or mesomelia, radiohumeral fusion, rib abnormalities, anomalies of the external genitalia and a potter-like facies. The syndrome has been described in three infants (one pair of sibs and an unrelated case), all of whom died shortly after birth from respiratory distress resulting from pulmonary hypoplasia and oligohydramnios caused by renal dysplasia. The mode of transmission appears to be autosomal recessive.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Renal dysplasia-limb defects syndrome
Dysplazja nerek - mezomelia - zrost kości promieniowej i ramiennej
Dysplazja nerek - wady kończyn
Renal dysplasia-mesomelia-radiohumeral fusion syndrome

Kod ORPHA
3404

Kod OMIM
266910

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet