Autosomalna recesywna wielotorbielowatość nerek Kod Orpha: 731 Kod OMIM: 617610

Opis choroby *

Definicja
A rare, genetic hepatorenal fibrocystic syndrome characterized by cystic dilatation and ectasia of renal collecting tubules, and a ductal plate malformation of the liver resulting in congenital hepatic fibrosis. Clinical presentation, whilst typically in utero or at birth, is variable and in the most severe cases includes Potter-sequence, oligohydramnios, pulmonary hypoplasia, and massively enlarged echogenic kidneys.

Dane

Klasyfikacja
Choroba

Synonimy
AR-PKD
AR-PKD

Kod ORPHA
731

Kod OMIM
617610

Kod ICD10
Q61.1

Kod ICD11
GB81

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.