Opis choroby *

Definicja

Renal-hepatic-pancreatic dysplasia is a rare, genetic, developmental defect during embryogenesis syndrome characterized by the triad of pancreatic fibrosis (and cysts, with a reduction of parenchymal tissue), renal dysplasia (with peripheral cortical cysts, primitive collecting ducts, glomerular cysts and metaplastic cartilage) and hepatic dysgenesis (enlarged portal areas containing numerous elongated binary profiles with a tendancy to perilobular fibrosis). Situs abnormalities, skeletal anomalies and anencephaly have also been associated. Patients that survive the neonatal period present renal insufficiency, chronic jaundice and insulin-dependent diabetes.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ivemark II syndrome
Zespół Ivemarka II
Renohepaticopancreatic dysplasia

Kod ORPHA
294415

Kod OMIM
615415

Kod ICD10
Q45.8

Kod ICD11
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*Źródło

orphanet