DISEASE:
Renal-hepatic-pancreatic dysplasia

NAME:	Renal-hepatic-pancreatic dysplasia
DESCRIPTION:	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.
ORPHACODE:	294415
SYNONYMS:	Ivemark II syndrome Renohepaticopancreatic dysplasia
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	NPHP3 NEK8
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RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Ciliopathy (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- NIMA related kinase 8
- nephrocystin 3

Related Gene Panels

- Ciliopathy (120 genes) - UGent
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG

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