DISEASE:
Autosomal recessive polycystic kidney disease

NAME:	Autosomal recessive polycystic kidney disease
DESCRIPTION:	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.
ORPHACODE:	731
SYNONYMS:	AR-PKD
XREF(S):	Orphanet MeSH MedDRA ICD-10 OMIM OMIM
ANALYTE(S):	DZIP1L PKHD1
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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)
- Hepatorenal disorders (gene panel)
- Inherited Kidney Diseases (Gene Panel)

Related Laboratories

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

Related Analytes

- DAZ interacting zinc finger protein 1 like
- PKHD1 ciliary IPT domain containing fibrocystin/polyductin

Related Gene Panels

- Cholestasis (40 genes) - UCL
- Ciliopathy (120 genes) - UGent

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Hepatorenal disorders (13 genes) - UCL
- Panel Nephro-ULG-V1
