DISEASE:
NPHP3-related Meckel-like syndrome

NAME:	NPHP3-related Meckel-like syndrome
DESCRIPTION:	NPHP3-related Meckel-like syndrome is a rare, genetic, syndromic renal malformation characterized by cystic renal dysplasia with or without prenatal oligohydramnios, central nervous system abnormalities (commonly Dandy-Walker malformation), congenital hepatic fibrosis, and absence of polydactyly.
ORPHACODE:	3032
SYNONYMS:	Goldston syndrome Meckel syndrome type 7 Meckel-like syndrome type 1 Renal-hepatic-pancreatic dysplasia-Dandy-Walker cysts syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	NPHP3
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

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

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- nephrocystin 3

Related Gene Panels

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