DISEASE:
HNF1B-related autosomal dominant tubulointerstitial kidney disease

NAME:	HNF1B-related autosomal dominant tubulointerstitial kidney disease
DESCRIPTION:	A form of autosomal dominant tubulointerstitial kidney disease (ADTKD) due to variants in or whole gene deletions of HNF1B, which is characterized by chronic tubulo-interstitial nephritis, that manifests with nonsignificant urinalysis and slowly progressive renal failure. It can be associated with cystic kidney dysplasia, early onset diabetes and extrarenal manifestations.
ORPHACODE:	93111
SYNONYMS:	ADTKD-HNF1B HNF1B-MODY HNF1B-related nephropathy MODY5 Maturity-onset diabetes of the young type 5 RCAD syndrome Renal cysts and diabetes syndrome Renal dysfunction-early-onset diabetes syndrome
XREF(S):	Orphanet OMIM MeSH ICD-10

ANALYTE(S):	HNF1B
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/3139

RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Hepatorenal disorders (gene panel)
- Renal cysts and diabetes syndrome
- Renal or urinary tract malformation (CAKUT) (gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- HNF1 homeobox B

Related Gene Panels

- Cakut (congenital anomalies of the kidney and urinary tract-1) (69 genes) - IPG
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG