DISEASE:
UMOD-related autosomal dominant tubulointerstitial kidney disease

NAME:	UMOD-related autosomal dominant tubulointerstitial kidney disease
DESCRIPTION:	A form of autosomal dominant tubulointerstitial kidney disease (ADTKD) due to UMOD mutations that is clinically characterized by bland urinalysis (absence of blood or protein in the urine), chronic kidney disease (CKD) leading to end-stage kidney disease (ESKD) between 20 and 80 years, and gout occurring in 50% of affected individuals.
ORPHACODE:	88950
SYNONYMS:	ADTKD-UMOD Familial juvenile hyperuricemic nephropathy type 1 MCKD2 Medullary cystic kidney disease type 2 UMOD-related ADTKD Uromodulin-associated kidney disease
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	UMOD
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Inherited Kidney Diseases (Gene Panel)
- Renal or urinary tract malformation (CAKUT) (gene panel)
- Tubulopathy (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- uromodulin

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
- Panel Nephro-ULG-V1
- Tubulopathy/Nephrolithiasis (106 genes) - IPG