DISEASE:
MUC1-related autosomal dominant tubulointerstitial kidney disease

NAME:	MUC1-related autosomal dominant tubulointerstitial kidney disease
DESCRIPTION:	A rare autosomal dominant tubulointerstitial kidney (ADTKD) disease due to MUC1 mutations characterized clinically by a bland urinalysis (absence of blood or protein in the urine), and chronic kidney disease leading to end-stage kidney disease (ESKD) between 20 and 80 years.
ORPHACODE:	88949
SYNONYMS:	ADTKD-MUC1 MCKD1 MUC1-related medullary cystic kidney disease MUCI-related ADTKD Medullary cystic kidney disease type 1
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MUC1
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- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

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- Panel Nephro-ULG-V1