DISEASE:
Karyomegalic interstitial nephritis

NAME:	Karyomegalic interstitial nephritis
DESCRIPTION:	A rare, genetic renal disease characterized by slowly progressive, chronic, tubulointerstitial nephritis, leading to end-stage renal disease before the age of 50 years, manifesting with mild proteinuria, glucosuria and, occasionally, urinary sediment abnormalities (mainly hematuria). Mild extrarenal manifestations, such as recurrent upper respiratory tract infections and abnormal liver function tests, may be associated. Renal biopsy reveals severe, chronic, interstitial fibrosis and tubular changes, as well as hallmark karyomegalic tubular epithelial cells which line the proximal and distal tubules and have enlarged, hyperchromatic nuclei.
ORPHACODE:	401996
SYNONYMS:	KIN Systemic karyomegaly
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FAN1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2288

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

- FANCD2 and FANCI associated nuclease 1

Related Gene Panels

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