# Tubulointerstitial diseases

THE GENOMIC INSTITUTE

## Step 1

**Document the following clinical information to assist in phenotyping:**

- Kidney function
- Blood biochemistry
- Urine analysis
- Kidney imaging
- Kidney biopsy, where indicated
- Extrarenal features
- Family history of kidney disease, including kidney phenotype

**For patients with a negative or unknown family history:**
If possible, assess the phenotype of both parents with kidney imaging, urine analysis, and kidney function testing.

**For patients with a positive family history:**
If a family member has had genetic testing and a disease-causing variant has been identified:

- Do not proceed to genomic testing
- Refer to Genetic Health Queensland to discuss targeted confirmatory genetic testing

**Consider genomic testing if there are any of the following indications for testing:**

- Diagnostic uncertainty
- Genotype-specific management
- Family planning
- Risk clarification for family
- Transplant planning (particularly if donor is a blood relative)

## Step 2

**The following are the key clinical diagnostic features from investigations:**

Urine

- Bland

Imaging

- Normal
- Microcysts
- Medullary cysts
- Increased echogenicity
- Decreased corticomedullary differentiation

Biopsy

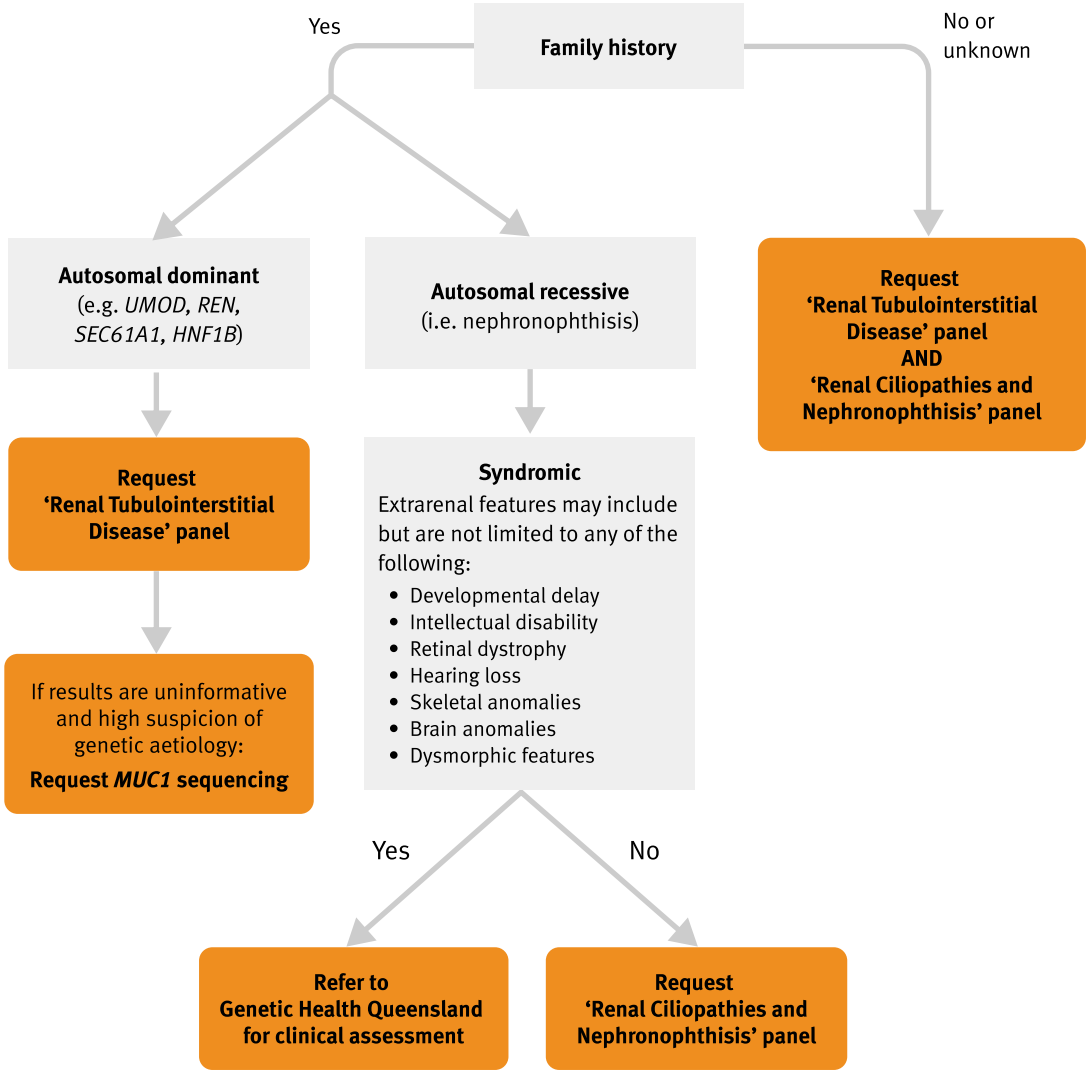
- Tubular atrophy
- Interstitial fibrosis

Blood

- High urate
- Low $Mg^{2+}$

## Step 3

**The following pathway guides genomic testing based on family history, suspected inheritance pattern, and extrarenal features.**






v1 Effective: June 2022

**For further specialist advice, email Renal Genetics MDT at Renal_Genetics_MDT@health.qld.gov.au**