Cystic diseases

THE GENOMIC INSTITUTE

Step 1

Document the following clinical information to assist in phenotyping:

- Size of kidneys
- Location, number, and size of cysts
- Cysts in other organs
- Kidney function
- Blood biochemistry
- 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/abdominal imaging 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 pathway guides genomic testing based on suspected clinical diagnosis. Key clinical features for each diagnosis are listed.

Autosomal Dominant Polycystic Kidney Disease (ADPKD)	• Enlarged kidneys • >10 kidney cysts, with family history • Liver cysts	→ **Request 'Renal Macrocystic Disease' panel** Although the same genomic test is requested for these diseases, detailed phenotype information is crucial for analysing and interpreting the genomic test.
Atypical cystic disease (e.g. *GANAB, DNAJB11*)	• Variable kidney sizes • Variable kidney cysts • Liver cysts	→ **Request 'Renal Macrocystic Disease' panel**
Autosomal Recessive Polycystic Kidney Disease (ARPKD)	• Kidney cysts • Increased echogenicity • Decreased corticomedullary differentiation • Hepatic fibrosis	→ **Request 'Renal Macrocystic Disease' panel**
Renal Cysts and Diabetes (RCAD)	• Kidney cysts • Tubulointerstitial kidney disease • CAKUT • Diabetes • Low Mg^{2+} • Bicornuate uterus	For paediatric patients: **Request chromosome microarray** If result is uninformative, proceed to request panel. → **Request 'Renal Macrocystic Disease' panel**
Nephronophthisis (NPHP)	• Kidney cysts (microcysts) • Increased echogenicity • Decreased corticomedullary differentiation • Tubular atrophy and interstitial fibrosis • Extrarenal features	→ **Go to 'Tubulointerstitial diseases'**

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