Glomerular diseases

THE GENOMIC INSTITUTE

Step 1

Document the following clinical information to assist in phenotyping:

- Kidney function
- Blood biochemistry
- Urine analysis, with serial urine PCR +/- ACR
- 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 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

- Haematuria
- Proteinuria

Biopsy (light and electron microscopy)

- Thin Glomerular Basement Membrane (GBM)
- Thickening and/or splitting of GBM
- Lamellation of GBM
- Podocyte effacement
- Focal Segmental Glomerular Sclerosis (FSGS)

Extrarenal features

- Sensorineural hearing loss
- Ophthalmology findings
- Neuropathy
- Other

Step 3

The following pathway guides genomic testing based on urine analysis findings.

Haematuria only (microscopic)	**Request ‘Haematuria_Alport’ panel**
Haematuria + Proteinuria	**Request ‘Renal Glomerular Disease_SuperPanel’ panel**
Proteinuria only +/- Nephrotic syndrome	**Request ‘Proteinuria’ panel**
Low molecular weight proteinuria	**Go to Tubulopathies**

v1 Effective: June 2022

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