Genetic Kidney Disease (GKD) Decision Aid

The GKD decision aid guides nephrologists to request genomic testing to diagnose GKD with a monogenic aetiology.

About this guide

Detailed phenotype information is crucial for selecting the correct genetic test, analysing the genetic test, and interpreting the genetic test.

Designed for common GKD groups, this decision aid includes specific diagnostic features, indications for genetic testing, and the recommendations for genomic testing for relevant diseases.

It also includes the mandatory steps for requesting genomic testing, including clinical phenotype details, gene panel selection, consent for testing, and a guide to interpreting results.

For patients with a negative or unknown family history:

If possible, assess the phenotype of both parents with investigations relevant to GKD group.

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

How to use this guide

1 Determine if any of the following are present to suggest possible GKD:

- Family history of kidney disease
- Young age of presentation with kidney disease
- Atypical presentation of kidney disease
- Diagnostic features of specific GKD identified on investigations

2 Document the following clinical information to assist in phenotyping:

- Age and symptoms at presentation
- Disease course
- Trend of kidney function
- Type of renal replacement therapy and age of kidney failure
- Extrarenal features
- Individuals in family with kidney disease, including kidney phenotype and age of kidney failure

3 Request the following relevant investigations to assist in phenotyping:

- Kidney function
- Blood biochemistry, with or without urine electrolytes
- Urine analysis, with serial urine protein to creatinine ratio (PCR) +/- albumin to creatinine ratio (ACR)
- Kidney or abdominal imaging
- Kidney biopsy, including electron microscopy

4 Request the appropriate genetic test based on suspected GKD

Contents

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

v1 | Effective: June 2022

Queensland Genomics | THE GENOMIC INSTITUTE | Genetic Health Queensland | Clinical Excellence Queensland | Queensland Government