Renal mainstream genetic testing protocol

PRE-TEST

Eligibility: meets GREEN criteria (Refer to Traffic Light Tool)

- ESKD<=50 with unclear cause
- Multiple renal cysts
- Suspected Alport syndrome or thin basement membrane nephropathy
- >6 months haematuria with no obvious cause
- A specific gene associated with kidney disease is suspected

N.B Some AMBER patients may be suitable for Mainstream pathway following discussion with Renal Genetic Counsellor or at central Kidney Genomics MDT meeting.

YES

Action points:

- Provide genetic test counselling to patient.
- Complete consent form with patient and upload to patient file
- Provide patient with genetic testing information sheet.
- Complete test request (online or paper request). Patient to attend blood collection.

Patient not eligible for mainstream pathway but genetics involvement indicated.

NO

Refer to Renal Genetics clinic

YES

Is further discussion needed prior to consent?

RESULTS

Result sent to requesting doctor (3-4 months from blood collection)

Recommended: Discuss results at Renal Genetics Mainstreaming MDT meeting (held via Zoom). Email case details to kidneygenetics@monashhealth.org.

Result (and any treatment and familial implications) can be discussed with patient by requesting doctor in renal clinic prior to referral to a renal genetics clinic. Provide patient with copy of the report.

NO GENETIC CAUSE IDENTIFIED **(negative)**

D. GENETIC CAUSE IDENTIFIED (positive)

FOLLOWUP

A. MONOGENIC CAUSE NO LONGER SUSPECTED

- No formal genetics follow-up is required (unless you have specific questions).
- Provide patient with negative results information sheet

B. ADDITIONAL TESTING TO BE CONSIDERED

1. Discuss at Central Renal MDT

Examples of indications to consider further testing:

- 2 or more family members affected
- Strong clinical suspicion of a specific genetic condition

C. VARIANT OF UNKNOWN SIGNIFICANCE (VUS)

1. Review VUS at central Renal Genetics MDT.

Depending on outcome of review at MDT:

- No follow up required. Provide patient with VUS results information sheet; or
- Refer to Renal Genetics Clinic

D. REFER TO RENAL GENETICS CLINIC

For further discussion regarding:

- Management implications/ clinical trials
- Genetic counselling
- Inheritance
- Identifying at-risk relatives
- Reproductive planning (patient or relatives)
- Emotional adjustment
- Facilitating testing of family members

For questions or support, contact your local genetic counsellor
email kidneygenomics@monashhealth.org

Monash Health · Austin Health · The Royal Melbourne Hospital · The Royal Children's Hospital Melbourne

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