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Decision Aid for Nephrologists:
Choosing the right test for my patient

Patient with suspected Genetic Kidney Disease (GKD)
Has the patient or family members had any previous genetic testing?

GKD risk factors
One or more of the following:
- Young age (<35) of presentation
- Family history of kidney disease
- Parental consanguinity
- Persistent haematuria >6months, no obvious cause
- CKD with bland urine*
- ESKD <50 with unclear aetiology

*If other causes excluded (eg hypertension)

Y → Obtain family test report and further information via **Genetic Counsellor support line**

N → Does the patient have any of the following?

Haematuria

Specific features of Alport syndrome? [1]

- **Y** → Test using **Alport gene panel** (MBS item 73298)
- **N** → Test using **Kidneyome super panel** [2] (MBS item 73402)

Multiple renal cysts
See **cystic pathway** for more info.

Other features (glomerular haematuria/ proteinuria/extrarenal) present?

- **Y** → Features of renal cysts and diabetes syndrome/ HNF1B? [3]
  - **Y** → 1. Test using Microarray (MBS item 73292) 2. If microarray result is non-diagnostic, the test using the WES[4] (MBS item 73402)
  - **N** → Test using Kidneyome super panel [2] (MBS item 73402)
- **N** → Test using **Cystic Kidney Disease super panel** (MBS item 73401)

Any findings from Microarray? Contact Genetic Counsellor support line to discuss (MBS item 73292)

Other:

Discuss at Multidisciplinary Team Meeting or call GC support line.

Contact the **Genetic Counsellor support line** for any questions about this decision aid.
Visit **kidneygenomics.org.au**

Melbourne Genomics Health Alliance

This resource was produced as part of the Scaling Up Genomics in Nephrology Project, June 2023.

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

About the Decision Aid

This tool was based on current available evidence and guidelines and after discussion between an expert working group comprising of nephrologists, geneticists, and genetic counsellors. A small number of patients may fall into more than one category. Contact your local genetic counsellor if you need clarification or have any feedback on this tool. Visit **kidneygenomics.org.au** for more information and contact details for the Kidney Genomics Multidisciplinary Team and the Genetic Counsellor support line.

Genetic Counsellor support line

The Genetic Counsellor support line may help to identify if there is a genomic result in the family for which cascade testing[5] (the process of offering genetic testing to at-risk blood relates of individuals who have been identified with specific genetic mutations) can occur.

Footnotes

1. Specific features of Alport syndrome include one of the following: high-tone sensorineural hearing loss, eye signs such as perimacular flecks or anterior lenticonus.
2. Other heritable kidney disease pathway includes testing for all genes associated with kidney disease (the Kidneyome) including Alport syndrome and cystic disease.
3. Features of HNF1B/RCAD (renal cysts and diabetes syndrome) include but are not limited to: renal cysts, other structural renal and/or genital tract anomalies, electrolyte abnormalities, abnormal liver function, early-onset diabetes mellitus and gout (Clissold et al., 2015).
4. WES = whole exome sequencing
5. Indication for cascade testing: the patient has a first-degree relative with a known monogenic cause of kidney disease identified by genetic testing, requested by a clinical geneticist; or a specialist providing professional genetic counselling services.

References

Clissold, R. L., Hamilton, A. J., Hattersley, A. T., Ellard, S., & Bingham, C. (2015). HNF1B-associated renal and extra-renal disease-an expanding clinical spectrum. *Nat Rev Nephrol, 11*(2), 102-112. https://doi.org/10.1038/nrneph.2014.232

Cystic pathway

Individuals with multiple kidney cysts (excluding those patients with suspected acquired cystic kidney disease) (Park et al., 2021).

- Individuals who meet ultrasound criteria for ADPKD but have no family history of cystic kidney disease.
- Individuals who have a clinical diagnosis of ADPKD where a genomic diagnosis will potentially benefit the individual being tested or their family member.

Examples for testing include reproductive planning, identification of at-risk relatives for prognosis/donor selection/screening, where there are atypical features (e.g. size of kidneys or kidney function decline).

Acquired cystic kidney disease should be suspected if there are ≥ 3 cysts in each kidney in a patient with CKD and small or normal sized kidneys. If unclear, screening family members with ultrasound may help to differentiate (Liu et al., 2000; Narasimhan et al., 1986; Rahbari-Oskoui & O'Neill, 2017)

References

Liu, J. S., Ishikawa, I., & Horiguchi, T. (2000). Incidence of acquired renal cysts in biopsy specimens. *Nephron, 84*(2), 142-147. https://doi.org/10.1159/000045562

Rahbari-Oskoui, F., & O'Neill, W. C. (2017). Diagnosis and Management of Acquired Cystic Kidney Disease and Renal Tumors in ESRD Patients. *Semin Dial, 30*(4), 373-379. https://doi.org/10.1111/sdi.12605

Narasimhan, N., Golper, T. A., Wolfson, M., Rahatzad, M., & Bennett, W. M. (1986). Clinical characteristics and diagnostic considerations in acquired renal cystic disease. *Kidney Int, 30*(5), 748-752. https://doi.org/10.1038/ki.1986.251

Park, H. C., Ryu, H., Kim, Y. C., Ahn, C., Lee, K. B., Kim, Y. H., Kim, Y., Han, S., Kim, Y., Bae, E. H., Ma, S. K., Kang, H. G., Ahn, Y. H., Park, E., Jeong, K., Lee, J., Choi, J., Oh, K. H., & Oh, Y. K. (2021). Genetic identification of inherited cystic kidney diseases for implementing precision medicine: a study protocol for a 3-year prospective multicenter cohort study. *BMC Nephrol, 22*(1), 2. https://doi.org/10.1186/s12882-020-02207-8