

## Supplementary Material

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## **Supplementary Appendix S1: Details of international survey methodology**

### **Participants**

Forty-five key kidney genetics clinic leads were identified through literature review, professional networks, and prior collaborations. Additional participants were recruited via snowball sampling, whereby existing participants nominated other relevant clinic leads.

### **Survey Development, Structure, and Content**

A 42-item survey instrument was developed using Google Forms, informed by pilot scoping review findings and expert consultation. The survey captured information across several key domains relevant to this analysis, including participant demographics, clinic characteristics and structure, patient populations and referral pathways, genetic counselling and testing practices, cascade testing and family management, and service funding models.

### **Survey Administration and Data Collection**

Survey invitations were distributed via personalized email, with up to three reminder emails sent at monthly intervals to non-respondents. Participation was voluntary. Formal ethics approval was not required as this stakeholder engagement was conceived and carried out within scoping review methodology. Completion of the survey following the email invitation was considered implied informed consent.

### **Data Analysis**

Survey responses were exported into Microsoft Excel for data cleaning and analysis. Descriptive statistics, including frequency counts and proportions, were used to summarize clinic characteristics and service delivery practices.

## Supplementary Table S1: PRISMA-ScR Checklist

### Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) Checklist

| SECTION                                               | ITEM | PRISMA-ScR CHECKLIST ITEM                                                                                                                                                                                                                                                                                  | REPORTED ON PAGE # |
|-------------------------------------------------------|------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------|
| <b>TITLE</b>                                          |      |                                                                                                                                                                                                                                                                                                            |                    |
| Title                                                 | 1    | Identify the report as a scoping review.                                                                                                                                                                                                                                                                   | 1                  |
| <b>ABSTRACT</b>                                       |      |                                                                                                                                                                                                                                                                                                            |                    |
| Structured summary                                    | 2    | Provide a structured summary that includes (as applicable): background, objectives, eligibility criteria, sources of evidence, charting methods, results, and conclusions that relate to the review questions and objectives.                                                                              | 3                  |
| <b>INTRODUCTION</b>                                   |      |                                                                                                                                                                                                                                                                                                            |                    |
| Rationale                                             | 3    | Describe the rationale for the review in the context of what is already known. Explain why the review questions/objectives lend themselves to a scoping review approach.                                                                                                                                   | 4                  |
| Objectives                                            | 4    | Provide an explicit statement of the questions and objectives being addressed with reference to their key elements (e.g., population or participants, concepts, and context) or other relevant key elements used to conceptualize the review questions and/or objectives.                                  | 4                  |
| <b>METHODS</b>                                        |      |                                                                                                                                                                                                                                                                                                            |                    |
| Protocol and registration                             | 5    | Indicate whether a review protocol exists; state if and where it can be accessed (e.g., a Web address); and if available, provide registration information, including the registration number.                                                                                                             | 5                  |
| Eligibility criteria                                  | 6    | Specify characteristics of the sources of evidence used as eligibility criteria (e.g., years considered, language, and publication status), and provide a rationale.                                                                                                                                       | 5                  |
| Information sources*                                  | 7    | Describe all information sources in the search (e.g., databases with dates of coverage and contact with authors to identify additional sources), as well as the date the most recent search was executed.                                                                                                  | 5                  |
| Search                                                | 8    | Present the full electronic search strategy for at least 1 database, including any limits used, such that it could be repeated.                                                                                                                                                                            | 5                  |
| Selection of sources of evidence†                     | 9    | State the process for selecting sources of evidence (i.e., screening and eligibility) included in the scoping review.                                                                                                                                                                                      | 5                  |
| Data charting process‡                                | 10   | Describe the methods of charting data from the included sources of evidence (e.g., calibrated forms or forms that have been tested by the team before their use, and whether data charting was done independently or in duplicate) and any processes for obtaining and confirming data from investigators. | 5-6                |
| Data items                                            | 11   | List and define all variables for which data were sought and any assumptions and simplifications made.                                                                                                                                                                                                     | 5-6                |
| Critical appraisal of individual sources of evidence§ | 12   | If done, provide a rationale for conducting a critical appraisal of included sources of evidence; describe the methods used and how this information was used in any data synthesis (if appropriate).                                                                                                      | NA                 |
| Synthesis of results                                  | 13   | Describe the methods of handling and summarizing the data that were charted.                                                                                                                                                                                                                               | 6-7                |

| SECTION                                       | ITEM | PRISMA-ScR CHECKLIST ITEM                                                                                                                                                                       | REPORTED ON PAGE # |
|-----------------------------------------------|------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------|
| <b>RESULTS</b>                                |      |                                                                                                                                                                                                 |                    |
| Selection of sources of evidence              | 14   | Give numbers of sources of evidence screened, assessed for eligibility, and included in the review, with reasons for exclusions at each stage, ideally using a flow diagram.                    | 8                  |
| Characteristics of sources of evidence        | 15   | For each source of evidence, present characteristics for which data were charted and provide the citations.                                                                                     | 26-27              |
| Critical appraisal within sources of evidence | 16   | If done, present data on critical appraisal of included sources of evidence (see item 12).                                                                                                      | NA                 |
| Results of individual sources of evidence     | 17   | For each included source of evidence, present the relevant data that were charted that relate to the review questions and objectives.                                                           | 8-11               |
| Synthesis of results                          | 18   | Summarize and/or present the charting results as they relate to the review questions and objectives.                                                                                            | 8-12               |
| <b>DISCUSSION</b>                             |      |                                                                                                                                                                                                 |                    |
| Summary of evidence                           | 19   | Summarize the main results (including an overview of concepts, themes, and types of evidence available), link to the review questions and objectives, and consider the relevance to key groups. | 13-16              |
| Limitations                                   | 20   | Discuss the limitations of the scoping review process.                                                                                                                                          | 16                 |
| Conclusions                                   | 21   | Provide a general interpretation of the results with respect to the review questions and objectives, as well as potential implications and/or next steps.                                       | 16                 |
| <b>FUNDING</b>                                |      |                                                                                                                                                                                                 |                    |
| Funding                                       | 22   | Describe sources of funding for the included sources of evidence, as well as sources of funding for the scoping review. Describe the role of the funders of the scoping review.                 | 17                 |

JB1 = Joanna Briggs Institute; PRISMA-ScR = Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews.

\* Where *sources of evidence* (see second footnote) are compiled from, such as bibliographic databases, social media platforms, and Web sites.

† A more inclusive/heterogeneous term used to account for the different types of evidence or data sources (e.g., quantitative and/or qualitative research, expert opinion, and policy documents) that may be eligible in a scoping review as opposed to only studies. This is not to be confused with *information sources* (see first footnote).

‡ The frameworks by Arksey and O'Malley (6) and Levac and colleagues (7) and the JBI guidance (4, 5) refer to the process of data extraction in a scoping review as data charting.

§ The process of systematically examining research evidence to assess its validity, results, and relevance before using it to inform a decision. This term is used for items 12 and 19 instead of "risk of bias" (which is more applicable to systematic reviews of interventions) to include and acknowledge the various sources of evidence that may be used in a scoping review (e.g., quantitative and/or qualitative research, expert opinion, and policy document).

**Supplementary Table S2: Final codebook of extracted implementation determinants from included studies, with corresponding study reference, country, illustrative quotes, interpretation, and mapped CFIR domains and constructs**

| Study Author, Year      | Country | Quotes                                                                              | Interpretation                                                                                                                                                                 | Determinants (Barrier or Facilitator) | CFIR Domain | CFIR Construct                   |
|-------------------------|---------|-------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------|-------------|----------------------------------|
| Jayasinghe et al., 2024 | AUS     | Delay in referrals and uptake by nephrologist not engaged with kidney genetics      | This reflects a gap in the implementation team's capability, where limited engagement and understanding of kidney genetics among nephrologists delay referrals and test uptake | Barrier                               | Individuals | Implementation Team-Capability   |
|                         |         | Nephrologists are unsure whom to refer to                                           | Reflects limited knowledge among nephrologists about appropriate referral criteria or patient selection for genetic testing.                                                   | Barrier                               | Individuals | Implementation Team-Capability   |
|                         |         | Nephrologists uncertain of the benefit of referral                                  | Reflects a lack of understanding or skepticism among nephrologists about the clinical utility or value of referring patients for genetic assessment.                           | Barrier                               | Individuals | Implementation Team-Capability   |
|                         |         | Results difficult for nephrologists without specific training to interpret/apply    | Reflects limited capability among nephrologists to interpret or apply genetic results without specialized training, hindering clinical integration.                            | Barrier                               | Individuals | Implementation Team-Capability   |
|                         |         | Difficulty in understanding the implications of a variant of uncertain significance | Reflects limited knowledge among nephrologists of implications and management plans for variant of uncertain significance.                                                     | Barrier                               | Individuals | Implementation Team-Capability   |
|                         |         | Delay or failure of whom to clinically act on genetic findings                      | Lack of role clarity as who to act on genetic findings.                                                                                                                        | Barrier                               | Individuals | Implementation Team-Capability   |
|                         |         | Nephrologists did not know how to order tests or prioritize patients for testing    | Reflects limited knowledge among nephrologists about appropriate patient prioritization and for genetic testing and test ordering.                                             | Barrier                               | Individuals | Innovation Deliverers-Capability |
|                         |         | Clinicians are unsure of which genes to test, which method to use                   | Reflects limited knowledge among nephrologists about genes to be tested and types of genetic testing to use.                                                                   | Barrier                               | Individuals | Innovation Deliverers-Capability |
|                         |         | The logistics of testing are too complicated to incorporate into clinics            | Reflects the perceived difficulty of integrating genetic testing logistics into routine clinical workflows due to its multifaceted and time-consuming processes.               | Barrier                               | Innovation  | Innovation Complexity            |

|  |  |                                                                                    |                                                                                                                                                                |         |               |                                   |
|--|--|------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------|---------|---------------|-----------------------------------|
|  |  | Unclear recommendation to referrers regarding the clinical application of findings | Reflects lack of communication of management recommendation to referring nephrologists regarding the clinical utility of genetic results post genetic testing. | Barrier | Innovation    | Innovation Complexity             |
|  |  | Long turn-around time for sequencing and analysis                                  | Reflects long turn-around time for genetic test reports and results.                                                                                           | Barrier | Innovation    | Innovation Complexity             |
|  |  | Variation in access to and cost of testing                                         | Reflects the lack of universal access and affordable cost to genetic testing.                                                                                  | Barrier | Innovation    | Innovation cost                   |
|  |  | No consistent clinical genetics support                                            | Reflects lack of consistent geneticists/genetics counsellors support to implement the kidney genetics service.                                                 | Barrier | Inner Setting | Available Resources               |
|  |  | The remote location of the clinical geneticist                                     | Reflects lack of onsite geneticists support to implement the kidney genetics service.                                                                          | Barrier | Inner Setting | Available Resources               |
|  |  | Unable to access clinical geneticist during clinic                                 | Reflects lack of access to geneticists support during clinic review.                                                                                           | Barrier | Inner Setting | Available Resources               |
|  |  | Delay in return of results                                                         | Reflects long wait time to return of genetic results after genetic testing due to lack of genetic clinicians availability.                                     | Barrier | Inner Setting | Available Resources               |
|  |  | The inequity of referral for testing                                               | Reflects underlying cultural norms and practices that contribute to inconsistent and inequitable referral patterns for genetic testing.                        | Barrier | Inner Setting | Culture                           |
|  |  | Clinicians are unsure of which genes to test, which method to use                  | Reflects nephrologists have limited access to kidney genetics knowledge and information about genes to be tested and types of genetic testing to use.          | Barrier | Inner Setting | Access to Knowledge & Information |
|  |  | Variable hospital funding for tests                                                | Reflects lack of sufficient funding provided by the hospital to support genetic testing.                                                                       | Barrier | Inner Setting | Financing                         |
|  |  | Poor community and consumer understanding of genomic testing                       | Reflects lack of consumer understanding in the community on the importance of genomic testing.                                                                 | Barrier | Outer Setting | Local Attitudes                   |
|  |  | Patients and the general community are uncertain of the benefits of referral       | Reflects lack of perceived benefits of genetic assessment referral among patients and the general community.                                                   | Barrier | Outer Setting | Local Attitudes                   |
|  |  | The remote location of patients who may lack literacy                              | Reflects low genomic literacy among patients living in the remote location.                                                                                    | Barrier | Outer Setting | Local Attitudes                   |
|  |  | Missed appointments to clinics reduce sustainability                               | Reflects high genetics clinic patient default rate which impacts the long-term sustainability of genetics clinic.                                              | Barrier | Outer Setting | Local Attitudes                   |

|                   |     |                                                                                                                            |                                                                                                                                                       |         |                        |                                |
|-------------------|-----|----------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------|---------|------------------------|--------------------------------|
|                   |     | Variable access to testing between jurisdictions                                                                           | Reflects differences in policies, regulations, or funding structures across regions that influence access to genetic testing.                         | Barrier | Outer Setting          | Policies and Laws              |
|                   |     | No secure funding for the multidisciplinary team (MDT) model of care                                                       | Reflects lack of secure funding from the ministry of health/government to support the implementation of multidisciplinary kidney genetics care model. | Barrier | Outer Setting          | Financing                      |
|                   |     | Delay in sequencing                                                                                                        | Reflects long turn-around time from genetic sequencing to return of genetic results.                                                                  | Barrier | Implementation Process | Doing                          |
|                   |     | Results not received by the referring doctor                                                                               | Reflects the genetic results were often not received by the referring nephrologist.                                                                   | Barrier | Implementation Process | Doing                          |
| Best et al., 2023 | AUS | Lack of self-confidence among clinicians in applying genetic knowledge in practice                                         | Represents self-efficacy and perceived ability to apply genomics in practice.                                                                         | Barrier | Individuals            | Implementation Team-Capability |
|                   |     | Limited knowledge of genetics among clinicians                                                                             | Refers to building knowledge and understanding                                                                                                        | Barrier | Individuals            | Implementation Team-Capability |
|                   |     | Limited training opportunities to develop genomic competencies and limited clinical skills in applying genomic information | Refers to acquiring and applying practical skills for genomic integration.                                                                            | Barrier | Individuals            | Implementation Team-Capability |
|                   |     | Implementation hindered by incomplete knowledge or experience                                                              | Ongoing learning indicates limited capability due to gaps in knowledge or skills necessary to implement genomic services confidently.                 | Barrier | Individuals            | Implementation Team-Capability |
|                   |     | Lack of confidence in communicating genomic results to patients                                                            | Clinicians may feel underprepared to explain complex genomic results clearly to patients.                                                             | Barrier | Individuals            | Implementation Team-Capability |
|                   |     | Lack of belief in the value of genomics                                                                                    | Reflects internal belief in the value/importance of genomics (attitudinal factor).                                                                    | Barrier | Individuals            | Implementation Team-Motivation |
|                   |     | Lack of trust in others                                                                                                    | Lack of trust in colleagues and multidisciplinary team to enable collaboration.                                                                       | Barrier | Inner Setting          | Relational Connections         |
|                   |     | Lack of trust among interdisciplinary team members                                                                         | Trust gaps may limit collaboration and interprofessional cooperation.                                                                                 | Barrier | Inner Setting          | Relational Connections         |
|                   |     | Lack of role clarity among team members                                                                                    | Lack of clearly defined roles leads to confusion and limits team effectiveness.                                                                       | Barrier | Inner Setting          | Work Infrastructure            |

|                             |     |                                                                                            |                                                                                                                                    |         |                        |                                     |
|-----------------------------|-----|--------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------|---------|------------------------|-------------------------------------|
|                             |     | Complex bureaucratic process and institutional procedures slow genomic service integration | Administrative burdens or procedural complexity hinder smooth implementation.                                                      | Barrier | Inner Setting          | Work Infrastructure                 |
|                             |     | Lack of a supportive or enabling implementation climate                                    | Describes the lack of norms and values shaping readiness for genomic care                                                          | Barrier | Inner Setting          | Culture                             |
|                             |     | Lack of psychological safety to express uncertainty or ask questions                       | Limited psychological safety hinders open communication and help-seeking behavior.                                                 | Barrier | Inner Setting          | Culture                             |
|                             |     | Lack of readiness to implement/integrate genomic services                                  | Insufficient preparation suggests lack of training, planning, or resources                                                         | Barrier | Inner Setting          | Access to Knowledge and Information |
|                             |     | Perceived complexity of the genomic testing process                                        | Refers to enacting workflows for genomic testing and service delivery                                                              | Barrier | Implementation Process | Doing                               |
|                             |     | Challenges in keeping up with evolving genomic knowledge and skills                        | Ongoing learning and adjusting based on experience and feedback                                                                    | Barrier | Implementation Process | Reflecting and Evaluating           |
|                             |     | Limited opportunities to build collaborative relationships                                 | The need to develop relationships suggests low initial engagement or weak team cohesion.                                           | Barrier | Implementation Process | Engaging                            |
| Kneifati-Hayek et al., 2024 | USA | Limited experience                                                                         | Clinicians lack experience in genomics, limiting confidence and ability to implement effectively                                   | Barrier | Individuals            | Implementation Team-Capability      |
|                             |     | Concern for unintended psychoemotional harm to patient/family                              | Clinicians worry that genomic information could emotionally harm patients/families                                                 | Barrier | Individuals            | Implementation Team-Motivation      |
|                             |     | Lack of ancillary support/staff                                                            | Insufficient support staff (e.g., genetic counselors, admin) limits capacity for service delivery.                                 | Barrier | Inner Setting          | Work Infrastructure                 |
|                             |     | Limited access to educational resources                                                    | Lack of access to training and resources hinders clinician preparedness and implementation readiness.                              | Barrier | Inner Setting          | Access to Knowledge and Information |
|                             |     | Concern it may lead to discrimination to patient/family/community                          | Fears of social or insurance discrimination discourage referrals and testing                                                       | Barrier | Outer Setting          | Policies and Laws                   |
|                             |     | Cost/lack of insurance coverage for testing                                                | High costs and lack of insurance coverage are key external financial barriers to access.                                           | Barrier | Outer Setting          | Financing                           |
| Fernandez et al., 2024      | USA | Diagnosis of untreatable kidney disease in asymptomatic children                           | Clinicians may hesitate to order testing that reveals non-actionable diagnoses, fearing emotional burden on patients and families. | Barrier | Individuals            | Implementation Team-Motivation      |

|  |  |                                                                                |                                                                                                                                          |             |               |                                     |
|--|--|--------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------|-------------|---------------|-------------------------------------|
|  |  | Challenges related to testing teenagers                                        | Clinicians face ethical and developmental concerns when offering genetic testing to adolescents.                                         | Barrier     | Individuals   | Implementation Team-Motivation      |
|  |  | Causing stigma for the patient                                                 | Clinicians fear that a genetic label may result in social or psychological harm.                                                         | Barrier     | Individuals   | Implementation Team-Motivation      |
|  |  | Lack of treatment options                                                      | Clinicians may be less motivated to test when actionable treatments are lacking.                                                         | Barrier     | Individuals   | Implementation Team-Motivation      |
|  |  | Identification of unexpected secondary/incidental genetic findings             | Unexpected findings complicate clinical decision-making and counseling.                                                                  | Barrier     | Individuals   | Implementation Team-Motivation      |
|  |  | Interpretation of results                                                      | Clinicians may feel unprepared to interpret complex genetic data.                                                                        | Barrier     | Individuals   | Implementation Team-capability      |
|  |  | Selection of the appropriate genetic test                                      | Clinicians may lack confidence in choosing the most appropriate genetic test.                                                            | Barrier     | Individuals   | Implementation Team-capability      |
|  |  | Providing genetic counseling for the family                                    | Limited access to counseling resources and time makes providing comprehensive genetic counseling difficult.                              | Barrier     | Inner Setting | Available Resources                 |
|  |  | Time to incorporate genetic testing into clinical practice                     | having protected or sufficient time signals an enabling work infrastructure to implement new practices like genomics.                    | Facilitator | Inner Setting | Available Resources                 |
|  |  | Administrative logistics                                                       | Complex administrative workflows, approvals, or consent processes deter clinicians.                                                      | Barrier     | Inner Setting | Structural Characteristics          |
|  |  | Designated person or team to lead the effort                                   | Having a designated lead or team reflects the presence of structural support and leadership roles aligned with implementation efforts.   | Facilitator | Inner Setting | Work Infrastructure                 |
|  |  | Unique challenges surrounding family counseling                                | Navigating complex family dynamics and communication about heritable risk is difficult.                                                  | Barrier     | Inner Setting | Compatibility                       |
|  |  | Access to reliable information to care for their patients with genetic results | Reflects clinicians' access to and use of reliable knowledge resources or data, supporting their ability to act on genetic test results. | Facilitator | Inner Setting | Access to Knowledge and Information |
|  |  | Navigating cost of genetic testing for patients                                | Clinicians are concerned that high costs may limit access or cause inequity.                                                             | Barrier     | Outer Setting | Financing                           |
|  |  | Unaffordability of genetic testing to their patients                           | Clinicians see testing as financially inaccessible to some patients, even when clinically indicated                                      | Barrier     | Outer Setting | Financing                           |

|                         |     |                                                                                                         |                                                                                                                                              |             |                        |                                |
|-------------------------|-----|---------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------|-------------|------------------------|--------------------------------|
|                         |     | Out-of-pocket costs for patients                                                                        | Financial burden from testing deters clinicians from recommending it widely.                                                                 | Barrier     | Outer Setting          | Financing                      |
|                         |     | Concern for denial of life insurance                                                                    | Clinicians worry about the long-term implications of testing results on patients' insurability.                                              | Barrier     | Outer Setting          | Policies and Laws              |
|                         |     | Concern for denial of disability insurance                                                              | Genetic results may lead to future insurance discrimination, influencing clinician hesitancy.                                                | Barrier     | Outer Setting          | Policies and Laws              |
|                         |     | Concern for denial of health insurance                                                                  | Concerns about patient discrimination from health insurers following genetic diagnosis.                                                      | Barrier     | Outer Setting          | Policies and Laws              |
|                         |     | Issues of privacy and confidentiality                                                                   | Concerns about secure handling and sharing of sensitive genetic information.                                                                 | Barrier     | Outer Setting          | Local Attitudes                |
| Mallet et al., 2016     | AUS | Clinic model is flexible                                                                                | Model can be tailored to different patient populations or settings                                                                           | Facilitator | Innovation             | Adaptability                   |
|                         |     | Clinic model is inclusive and multidisciplinary                                                         | Clinic was deliberately designed to be inclusive and multidisciplinary                                                                       | Facilitator | Innovation             | Innovation Design              |
|                         |     | Clinic model improves patient diagnosis and care                                                        | Clinic model improves patient diagnosis and care compared to conventional model of care                                                      | Facilitator | Innovation             | Relative Advantage             |
| Jayasinghe et al., 2021 | AUS | Knowledge gaps among nephrologists surrounding genetics                                                 | Captures clinicians' current level of knowledge and skill related to genomics, which affects their ability to engage with the innovation     | Barrier     | Individuals            | Implementation Team-Capability |
|                         |     | Detailed phenotypic information for a more accurate phenotype-genotype correlation                      | Reflects clinicians' understanding that rich phenotype data improves diagnostic accuracy                                                     | Barrier     | Individuals            | Implementation Team-Capability |
|                         |     | Limitation of exome sequencing to detect mitochondrial, structural, non-coding variants                 | Technical limitations and the incomplete coverage of the genomic test itself.                                                                | Barrier     | Innovation             | Innovation Design              |
|                         |     | To perform chromosomal microarray analysis prior to clinical whole exome sequencing genomic test        | Reflects the ability to tailor or sequence genomic testing strategies to fit patient needs and testing goals.                                | Facilitator | Innovation             | Adaptability                   |
|                         |     | Slow turn-around time of 6 months for return of genomic results                                         | Reflects the organizational infrastructure and system processes that influence the efficiency of genomic service delivery.                   | Barrier     | Inner Setting          | Structural characteristics     |
|                         |     | Efficiency improved with improved experience among clinicians, variant prioritization, and MDT meetings | Captures how increasing familiarity, collaboration, and streamlined workflows enhance effective implementation of genomic testing practices. | Facilitator | Implementation Process | Doing                          |

|                        |         |                                                                                                                                                                                                                                    |                                                                                                                                                                                                                                             |             |               |                                  |
|------------------------|---------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------|---------------|----------------------------------|
| Alkanderi et al., 2017 | UK      | Lack of advanced genomic testing to confirm pathogenicity of VUS/unsolved cases                                                                                                                                                    | Limits confidence in interpreting and acting on VUS results.                                                                                                                                                                                | Barrier     | Innovation    | Innovation Design                |
|                        |         | Complexities of monogenic inherited diseases and the intricacies of genetic counselling and predicting long term prognosis for patients                                                                                            | Reflects the complexity of understanding genetic kidney disease, details of genetic counselling, and long-term prognosis prediction in clinical setting,                                                                                    | Barrier     | Innovation    | Complexity                       |
|                        |         | Cost of multidisciplinary kidney genetics clinic                                                                                                                                                                                   | Perceived financial burden limits feasibility of clinic setup or sustainability.                                                                                                                                                            | Barrier     | Inner Setting | Available Resources              |
| Robinson et al., 2019  | UK      | High-quality, tailored information supported patient informed decision-making                                                                                                                                                      | Patients reported that the quality of tailored information improved their understanding and confidence in decision-making                                                                                                                   | Facilitator | Individuals   | Innovation Recipients-Capability |
|                        |         | Specialist expertise in inherited renal disorders                                                                                                                                                                                  | Nephrologists with expertise in kidney genomic enhance the implementation of kidney genetics service                                                                                                                                        | Facilitator | Individuals   | Innovation Deliverers-Capability |
|                        |         | Range and continuity of care provided within one clinic                                                                                                                                                                            | clinic culture that prioritizes coordinated, seamless, and patient-centered care.                                                                                                                                                           | Facilitator | Inner Setting | Culture – Recipient-Centeredness |
|                        |         | Easy access to specialist advice, often via the specialist nurse, for patients, general practitioners and local physicians.                                                                                                        | Presence of effective formal and informal communication channels and knowledge flow across team members and external stakeholders                                                                                                           | Facilitator | Inner Setting | Communication                    |
| Elhassan et al., 2022  | Ireland | We also used specific clinical criteria to limit our population to a group of patients with a high risk of familial kidney disease.                                                                                                | Systematic identification of high-risk patients using clinical criteria reflects a recognized clinical need by clinicians for better-targeted genomic testing.                                                                              | Facilitator | Individuals   | Innovation Recipients-Need       |
|                        |         | The ability to identify <i>MUC1</i> variants was important for the evaluation of genetic kidney disease in our cohort, using both <i>MUC1</i> genetic sequencing and newer techniques to detect the mutant <i>MUC1</i> fs protein. | Offers a clear diagnostic benefit over standard sequencing approaches by addressing technically challenging regions (e.g. <i>MUC1</i> VNTR), thus increasing the diagnostic yield for autosomal dominant tubulointerstitial kidney disease. | Facilitator | Innovation    | Relative Advantage               |
|                        |         | A targeted gene panel, designed to achieve high coverage of <i>PKD1</i> and <i>PKD2</i> was utilised in patients with                                                                                                              | This targeted panel offers a high diagnostic yield (78.7%) in a well-defined patient group with suspected PKD, demonstrating its clear                                                                                                      | Facilitator | Innovation    | Relative Advantage               |

|                     |     |                                                                                                                                                                                                              |                                                                                                                                                        |             |               |                                          |
|---------------------|-----|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------|-------------|---------------|------------------------------------------|
|                     |     | an a priori diagnosis of polycystic kidney disease (PKD)                                                                                                                                                     | advantage over broader or less focused testing which may miss PKD pseudogenes                                                                          |             |               |                                          |
|                     |     | The use of whole exome sequencing (WES) or indeed whole genome sequencing (WGS) allows future-proofing of diagnostic tests, allowing for reanalysis of data as novel genes are discovered.                   | Perceived advantage of broader utility and adaptability of genomic data                                                                                | Facilitator | Innovation    | Relative Advantage                       |
|                     |     | Integrated collaboration between clinical geneticists and nephrologists                                                                                                                                      | Effective communication and collaboration within and across specialties.                                                                               | Facilitator | Inner Setting | Relational Connections and communication |
|                     |     | Licensed clinical geneticist or genetic counsellors were not involved in patient review                                                                                                                      | Indicates a resource gap in terms of specialized personnel necessary for high-quality genomic care delivery.                                           | Barrier     | Inner Setting | Available Resources                      |
| Thomas et al., 2020 | USA | Lack of understanding of the power and limitations of sequencing technology among nephrologists                                                                                                              | Refers to knowledge and skill gaps in the clinical workforce that hinder effective genomic integration                                                 | Barrier     | Individuals   | Innovation Deliverers-Capability         |
|                     |     | Many genetic disorders have an incomplete or overlapping phenotype and may phenocopy an acquired disease, thus causing a genetic disease to go unrecognized                                                  | Phenotypic overlap and diagnostic ambiguity increase perceived complexity, contributing to under-referral for genetic services.                        | Barrier     | Innovation    | Complexity                               |
|                     |     | The genetic test results, especially when scored based on ACMG criteria, can be confusing to the clinician when a series of VUS are reported in genes that may not even be relevant to the patient's disease | Highlights the inherent challenge in interpreting VUS, increasing the cognitive and workflow burden on clinicians.                                     | Barrier     | Innovation    | Complexity                               |
|                     |     | Use of comprehensive genetic test panel (264 genes)                                                                                                                                                          | A comprehensive gene panel reflects a well-designed and clinically relevant testing tool that enhances the diagnostic yield                            | Facilitator | Innovation    | Innovation Design                        |
|                     |     | Clinical genetic counsellor is a vital part of any genetics clinic                                                                                                                                           | The presence of a clinical genetic counsellor provides essential expertise and support, facilitating successful implementation of the genetics clinic. | Facilitator | Inner Setting | Available Resources                      |
|                     |     | Rapidly changing landscape of testing labs and capabilities makes                                                                                                                                            | Frequent changes in local lab availability, capabilities, and processes create uncertainty in                                                          | Barrier     | Outer Setting | Local Conditions                         |

|                        |     |                                                                                                                                                                                    |                                                                                                                                                           |             |               |                                                |
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|                        |     | it challenging for a clinician to determine the best testing approach, or how and where to have genetic testing done                                                               | accessing and choosing appropriate genetic testing services.                                                                                              |             |               |                                                |
|                        |     | Some insurance payers require preauthorization, which can again dampen a clinician's enthusiasm for ordering these tests                                                           | Reflects administrative and payer-related external policy barriers that delay or limit access to testing.                                                 | Barrier     | Outer Setting | Policies and Laws                              |
| Thomas et al., 2021    | USA | Physicians and genetic counselors are aware of the limitations of genetic testing by next-generation sequencing and to know when additional diagnostic techniques may be necessary | Reflects the importance of user understanding of testing limitations and complementary diagnostic strategies                                              | Facilitator | Individuals   | Implementation Team-Capability                 |
|                        |     | A systematic approach to genetic testing of living donor candidates                                                                                                                | Reflects a well-organized and clearly defined approach that supports effective implementation.                                                            | Facilitator | Innovation    | Innovation design                              |
|                        |     | Integration of a renal genetics service with a genetics counselor into the renal transplant clinic for efficient deployment of genetic testing                                     | Reflects the embedding of new roles and services into existing organizational structures to support implementation                                        | Facilitator | Inner Setting | Structural Characteristics-Work infrastructure |
|                        |     | The costs of genetic testing of recipient and donor candidates are covered by recipient insurance or recorded as part of the organ acquisition cost on the Medicare cost report    | Reflects availability of external reimbursement mechanisms that support implementation.                                                                   | Facilitator | Outer Setting | Financing                                      |
| Lundquist et al., 2020 | USA | A physician with an interest in inherited kidney disease                                                                                                                           | A physician's interest in inherited kidney disease reflects intrinsic motivation, which supports engagement and implementation success.                   | Facilitator | Individuals   | Implementation Team-Motivation                 |
|                        |     | Panel-based testing as an initial strategy to reduce cost and to expedite results.                                                                                                 | Panel-based testing offers a more efficient and cost-effective alternative to broader testing, improving timeliness and perceived value of the innovation | Facilitator | Innovation    | Relative advantage                             |

|                         |     |                                                                                                                                                                                                                                     |                                                                                                                                       |             |                        |                                                |
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|                         |     | We believe that initial screening with more affordable and focused panel-based testing, followed by more comprehensive WES in patients with a strong family history of disease and negative panel testing, is an effective approach | The structured, tiered testing strategy reflects a thoughtfully designed and purposeful implementation of the innovation.             | Facilitator | Innovation             | Innovation Design                              |
|                         |     | A part-time genetic counselor was an essential part of the clinic, counselling patients and serving as a liaison with the testing laboratory for sample coordination and insurance coverage                                         | The part-time genetic counsellor provided critical clinical and operational support, enhancing clinic functionality and coordination. | Facilitator | Inner Setting          | Available Resources                            |
|                         |     | Supportive administration                                                                                                                                                                                                           | Administrative support ensures access to necessary resources (e.g., funding, space, personnel) for implementation.                    | Facilitator | Inner Setting          | Available Resources                            |
|                         |     | These experiences underscore the successful use of a shared and centrally hired genetic counselor, as opposed to hiring a full-time genetic counselor for each clinic.                                                              | A shared, centrally hired genetic counselor reflects an efficient organizational structure that supports sustainable implementation.  | Facilitator | Inner Setting          | Structural Characteristics-Work infrastructure |
|                         |     | Widespread testing may remain cost prohibitive for some individuals without insurance coverage in other parts of the United States or internationally.                                                                              | The high cost of genetic testing limits accessibility for some populations, creating economic barriers to implementation.             | Barrier     | Outer Setting          | Financing                                      |
|                         |     | We have subsequently experienced an increase in referrals with increased awareness of the clinic.                                                                                                                                   | Increased awareness of the clinic reflects successful stakeholder engagement, leading to greater referral uptake.                     | Facilitator | Implementation Process | Engaging                                       |
| Amlie-Wolf et al., 2021 | USA | Eight families declined due to lack of interest in testing                                                                                                                                                                          | Indicates low perceived value or relevance of genetic testing among patients or families.                                             | Barrier     | Individuals            | Innovation Recipients-Motivation               |
|                         |     | Identifying the appropriate genetic testing is the first barrier, but other factors further complicate the process, such as insurance/cost, informed consent and coordination of testing, which can be challenging                  | This reflects the inherent complexity of the genetic testing process itself                                                           | Barrier     | Innovation             | Innovation Complexity                          |

|                            |     |                                                                                                                                                                                                                                                                           |                                                                                                                                                                                                                                                  |             |               |                        |
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|                            |     | to navigate, even for trained genetics providers.                                                                                                                                                                                                                         |                                                                                                                                                                                                                                                  |             |               |                        |
|                            |     | Nephrologist was involved in the testing, they are able to make recommendations for medical management changes more quickly than if the Medical Genetics Clinic had to order and interpret results and refer back to the referring provider for treatment and management. | This highlights the added benefit and efficiency of nephrologist-led testing, where direct involvement enables more rapid interpretation and action on genetic results compared to traditional referral-based pathways through genetics clinics. | Facilitator | Innovation    | Relative advantage     |
|                            |     | Genetic Stewardship Programme offers in person and phone consenting sessions to accommodate families who cannot come to the hospital for an in-person visit                                                                                                               | This demonstrates flexibility in how the innovation (genetic testing and consenting process) is delivered, allowing it to be tailored to meet the needs and circumstances of different families, which supports implementation.                  | Facilitator | Innovation    | Adaptability           |
|                            |     | The collaboration between Genetic Stewardship Programme and Nephrology overcomes these barriers by providing genetic testing to patients in a more effective manner                                                                                                       | Effective collaboration and strong working relationships between internal departments, enabling more efficient delivery of genetic testing.                                                                                                      | Facilitator | Inner Setting | Relational Connections |
|                            |     | Utilizing genetic counsellors in Nephrology has been fruitful in managing cascade testing for family members.                                                                                                                                                             | The integration of genetic counselors within Nephrology represents a valuable human resource that supports effective coordination and delivery of cascade testing                                                                                | Facilitator | Inner Setting | Available Resources    |
|                            |     | Eight families declined due to high expected out-of-pocket costs                                                                                                                                                                                                          | Reflects affordability issues deterring patients from pursuing testing                                                                                                                                                                           | Barrier     | Outer Setting | Financing              |
| Pinto e Vairo et al., 2021 | USA | Employing a more targeted research gene panel with higher coverage of the genes tested to detect copy number variation missed by exome sequencing and standard gene panels                                                                                                | This refers to a deliberate refinement in the design of the genetic testing approach—using a targeted panel with enhanced coverage to address known limitations of exome sequencing                                                              | Facilitator | Innovation    | Innovation Design      |
|                            |     | The incorporation of specialized genetic counsellors into our nephrology practice improved                                                                                                                                                                                | The integration of specialized genetic counsellors reflects the availability and deployment of key human resources within the                                                                                                                    | Facilitator | Inner Setting | Available Resources    |

|                         |     |                                                                                                                                                                                                                                                                                                                                                                                                                                     |                                                                                                                                                                                                                                                     |             |                        |                                 |
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|                         |     | access to genetic testing and provided a substantial impact in this patient population                                                                                                                                                                                                                                                                                                                                              | practice, directly enhancing access to genetic testing and improving service delivery.                                                                                                                                                              |             |                        |                                 |
|                         |     | Twenty individuals did not undergo genomic testing due to reimbursement issues                                                                                                                                                                                                                                                                                                                                                      | Reimbursement limitations are a financial policy issue external to the organization that directly affects patient access to genomic testing and hinders implementation efforts.                                                                     | Barrier     | Outer Setting          | Financing                       |
|                         |     | The Nephrology Genomics Board reviews case details with the referring nephrologist and interprets the genetic results relevant to the patient's phenotype. In addition to verifying eligibility and approving research activities, and providing guidance to clinical care teams regarding possible ancillary testing, treatments, and clinical trials, the Board promotes genomic education and outreach within the practice group | The Genomics Board facilitates engagement, clinical decision support, education, and leadership—all within the implementation process.                                                                                                              | Facilitator | Implementation Process | Engaging-Innovation deliverers  |
| Bekheirnia et al., 2021 | USA | Access to world-class pediatric nephrology and clinical genetics groups                                                                                                                                                                                                                                                                                                                                                             | This emphasizes the high level of clinical expertise, training, and competency of the implementation team, which supports effective delivery and integration of genetic services into practice.                                                     | Facilitator | Individuals            | Implementation Team-Capability  |
|                         |     | 14 families were not interested to pursue genetic testing                                                                                                                                                                                                                                                                                                                                                                           | Lack of interest from families reflects low motivation or perceived value of the innovation among the intended recipients, which can impede uptake and implementation.                                                                              | Barrier     | Individual             | Innovation Recipient-Motivation |
|                         |     | Broad genetic testing options were available in our center. Our testing approach utilized various combinations of targeted panels, chromosomal microarray analysis and exome sequencing. The ability to perform advanced clinical genetic testing for a large proportion of our patients                                                                                                                                            | The availability of a broad range of genetic testing options reflects the thoughtful design and comprehensive nature of the innovation itself, enhancing its ability to meet diverse clinical needs and improving its perceived quality and utility | Facilitator | Innovation             | Innovation Design               |

|                    |     |                                                                                                                                                                                                                                                |                                                                                                                                          |             |               |                                 |
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|                    |     | The diagnostic rate of CAKUT in this cohort is higher than expected from literature. This is likely due to stringent referral criteria that select syndromic patients.                                                                         | The tailored referral strategy improves diagnostic yield, showing the innovation performs better than typical approaches.                | Facilitator | Innovation    | Relative Advantage              |
|                    |     | We were not able to perform genetic testing for 28 individuals (14 because of insurance denial)                                                                                                                                                | Insurance denial is an external financial and policy-related barrier that limits access to genetic testing and hinders implementation.   | Barrier     | Outer Setting | Financing                       |
| Bogyo et al., 2022 | USA | Among the referrals where a new patient genetic consult was not scheduled, no contact with the patient (n=33) was the most cited barrier, followed by patient declined or lack of interest (n=15)                                              | Patients' lack of interest or decision to decline reflects a motivational barrier to engagement.                                         | Barrier     | Individual    | Innovation Recipient-Motivation |
|                    |     | The most common reason for test cancelation was due to patient non-compliance (i.e. not sending in a sample or completing test consents), these rates were similar regardless of insurance status or type                                      | Reflects a lack of patient follow-through that may relate to low motivation, understanding, or perceived value of testing.               | Barrier     | Individual    | Innovation Recipient-Motivation |
|                    |     | Though the clinic offered the ability for patients to self-refer, few of the referrals came directly from patients.                                                                                                                            | May reflect limited health literacy, understanding of the clinic's purpose, or confidence to self-initiate a referral.                   | Barrier     | Individual    | Innovation Recipient-Capability |
|                    |     | Providers were given many options regarding how to refer their patients, including messaging a clinic-specific email, Electronic Medical Record (EMR) referral, online referral form, phone call, or email to a clinic staff's personal email. | The referral process is tailored to meet provider needs and preferences, showing adaptability of the implementation to the local context | Facilitator | Innovation    | Adaptability                    |
|                    |     | The structure of the Kidney Genetics Clinic was created such that it provides flexibility to providers, offering multiple                                                                                                                      | Reflects intentional design of the clinic to allow customization and responsiveness to provider preferences and workflow needs           | Facilitator | Innovation    | Adaptability                    |

|  |  |                                                                                                                                                                                                          |                                                                                                                                                                |             |                        |                                       |
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|  |  | appointment types and points of entry into the clinic workflow.                                                                                                                                          |                                                                                                                                                                |             |                        |                                       |
|  |  | By continuing to utilize a combination of in-person appointments and telemedicine, the Kidney Genetics Clinic will help genetic services reach a broad geographic region and diverse patient population. | The hybrid delivery model demonstrates adaptability to different patient needs and geographic constraints.                                                     | Facilitator | Innovation             | Adaptability                          |
|  |  | A monthly multidisciplinary genetic variant sign-out meeting and educational series were established                                                                                                     | Regular multidisciplinary meetings promote strong team communication, shared understanding, and collaboration across specialties                               | Facilitator | Inner Setting          | Relational Connections                |
|  |  | Nephrologists and genetic counsellors have collaborated to create a comprehensive Kidney Genetics Clinic                                                                                                 | Demonstrates effective interdisciplinary collaboration and strong working relationships across roles.                                                          | Facilitator | Inner Setting          | Relational Connections                |
|  |  | Technology issues, and cost were rare but reported barriers to scheduling new patient genetic consults.                                                                                                  | Reflects limitations in infrastructure (e.g., digital access)                                                                                                  | Barrier     | Inner Setting          | Information Technology Infrastructure |
|  |  | Lack of professional state licensure                                                                                                                                                                     | Lack of licensure reflects regulatory and policy-level barriers outside the organization's control                                                             | Barrier     | Outer Setting          | Policies and Laws                     |
|  |  | Physician and genetic counsellor licensure needs to be a consideration in seeing patients located in other states.                                                                                       | State-based licensure restrictions create regulatory barriers that limit cross-jurisdictional service delivery.                                                | Barrier     | Outer Setting          | Policies and Laws                     |
|  |  | Technology issues, and cost, were rare but reported barriers to scheduling new patient genetic consults.                                                                                                 | Lack of funding to support genomic assessment in eligible patients                                                                                             | Barrier     | Outer Setting          | Financing                             |
|  |  | Referring clinical nephrologists were involved throughout this process.                                                                                                                                  | Demonstrates active engagement of key stakeholders (referring nephrologists) in the implementation process, fostering ownership, collaboration, and relevance. | Facilitator | Implementation Process | Engaging-Innovation Deliverers        |
|  |  | Familiarizing nephrologists to the research workflow and educating them on genetic topics commonly                                                                                                       | Offers regular, structured opportunities for nephrologists to build competencies in renal genetics                                                             | Facilitator | Implementation Process | Engaging-Innovation Deliverers        |

|                      |     |                                                                                                                                                                              |                                                                                                                                                                                              |             |               |                        |
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|                      |     | encountered in the clinic, facilitated the establishment and utilization of the Kidney Genetics Clinic.                                                                      |                                                                                                                                                                                              |             |               |                        |
| El Ters et al., 2023 | USA | Challenges exist in interpretation of results, especially when revealing a VUS that may be challenging                                                                       | VUS interpretation requires nuanced understanding and increases the difficulty of clinical decision-making.                                                                                  | Barrier     | Innovation    | Innovation Complexity  |
|                      |     | Staff needs to submit needed paperwork, and track results.                                                                                                                   | Staffing capacity and time required for administrative tasks may limit implementation.                                                                                                       | Barrier     | Inner Setting | Available Resources    |
|                      |     | These challenges can be, at least in part, alleviated by the support from the genetic counselor team, as well as by having a dedicated nephrology genetic focus group (NGB). | Access to a genetic counselor team and a focused expert group provides personnel and structural support for implementation.                                                                  | Facilitator | Inner Setting | Available Resources    |
|                      |     | The success in incorporating genomic testing in kidney transplant evaluation relies primarily on a multidisciplinary team approach                                           | A multidisciplinary team approach reflects strong interpersonal relationships, communication, and collaboration within the inner setting, which are essential for coordinated implementation | Facilitator | Inner Setting | Relational Connections |
|                      |     | Of those, 30 completed genetic testing, and 4 canceled—3 because of reimbursement issue and 1 because they withdrew consent.                                                 | Reimbursement issues are external systemic financial barriers.                                                                                                                               | Barrier     | Outer Setting | Financing              |
|                      |     | These challenges include cost of the testing/insurance coverage                                                                                                              | Cost and reimbursement are external factors that impact the feasibility and uptake of testing                                                                                                | Barrier     | Outer Setting | Financing              |
|                      |     | Turnaround time typically in the range of 6 to 8 week and sometimes longer depending on the financial approval process                                                       | Insurance policies like prior authorization delay access.                                                                                                                                    | Barrier     | Outer Setting | Policies and Laws      |
| Tan et al., 2023     | USA | A comprehensive pretest evaluation served as an initial screen for testing necessity thereby increasing overall diagnostic yield and cost-effectiveness                      | A well-designed pretest process enhances the appropriateness and efficiency of genomic evaluation                                                                                            | Facilitator | Innovation    | Innovation Design      |
|                      |     | We utilized a multi-tiered approach to diagnostic testing, with multigene panels most frequently utilized. The selection of multigene                                        | The testing strategy reflects a well-organized, thoughtful design that enhances usability and performance                                                                                    | Facilitator | Innovation    | Innovation Design      |

|                       |     |                                                                                                                                                                                                                                                                  |                                                                                                                                     |         |               |                   |
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|                       |     | panel was phenotype-driven and resulted in a high diagnostic rate in our center                                                                                                                                                                                  |                                                                                                                                     |         |               |                   |
|                       |     | Medicare had a significantly lower genetic testing approval rate compared to private insurance (33% vs 66%) and Medicaid (33% vs 70%)<br>Finally, denial by insurers for renal genetic testing is common and Medicare is most likely to decline testing coverage | Lower approval rates under Medicare reflect funding-related barriers that affect access to genomic services.                        | Barrier | Outer Setting | Financing         |
|                       |     | No cost testing offered by commercial vendors served as a valuable option for expanding access to patient populations with access barriers (lack of insurance coverage or inability to pay high copay)                                                           | Removing cost barriers enhanced equity and enabled testing in underserved populations                                               | Barrier | Outer Setting | Financing         |
|                       |     | Despite a potential overturn of initial denial of coverage, the appeal process inevitably deterred patients from proceeding. This generated administrative overhead and led to diagnostic delay                                                                  | The appeal process reflects payer-driven financial and policy barriers that limit timely access to genomic testing.                 | Barrier | Outer Setting | Policies and Laws |
| McFadden et al., 2024 | USA | The discrepancy likely reflects significant variation in referral patterns at different institutions and highlights a need for additional evidence to more accurately identify high risk patients who should undergo genetic testing.                            | Limited or inconsistent evidence for identifying high-risk patients results in variable referral practices and underuse of testing. | Barrier | Innovation    | Evidence-base     |
|                       |     | Genetic testing patterns and panels were not uniform, owing to a number of patients with testing performed through other clinics, often prior to being seen in our clinic.                                                                                       | Inconsistent test selection and workflows reduce standardization and affect implementation fidelity.                                | Barrier | Innovation    | Innovation Design |

|                    |     |                                                                                                                                                                                                                                                         |                                                                                                                        |             |               |                       |
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|                    |     | Multidisciplinary clinic to support patients with informatics, care coordination, trial and research access, and patient empowerment                                                                                                                    | The clinic provides infrastructure, personnel, and services that facilitate comprehensive care and access to research  | Facilitator | Inner Setting | Available Resources   |
| Zhang et al., 2024 | USA | We could not identify the clinical significance of certain variants with either likely pathogenic effect or unknown significance.                                                                                                                       | Ambiguous or uncertain variant interpretation increases clinical and cognitive burden, complicating decision-making.   | Barrier     | Innovation    | Innovation Complexity |
|                    |     | We did not have a genetic counsellor in the clinic to better educate our patients on their findings and alleviate any unnecessary anxiety                                                                                                               | Absence of essential personnel (genetic counsellor) constrained service delivery and patient support.                  | Barrier     | Inner Setting | Available Resources   |
|                    |     | Almost all of our patients did not have an out-of-pocket cost, as most of the insurance companies covered the co-pay.                                                                                                                                   | Adequate insurance coverage supports equitable access by removing cost-related barriers.                               | Facilitator | Outer Setting | Financing             |
|                    |     | Much of the testing was not delayed by coordinating insurance coverage. Moreover, there was no clinic administrative time spent addressing any insurance coverages or follow-up on payments once the order had been placed.                             | Smooth insurance processes and full coverage helped avoid common cost and administrative barriers.                     | Facilitator | Outer Setting | Policies and Laws     |
| Payne et al., 2024 | USA | Currently, there are no clear American Urology Association guidelines on when to use genetic testing for patients with nephrolithiasis, and thus the decision to refer to genetics is provider and institution dependent                                | The absence of guidelines reflects a weak evidence base, limiting confidence in when and how to apply genetic testing. | Barrier     | Innovation    | Evidence-base         |
|                    |     | Use of key referral criteria for genetics referral e.g. family history, multiple past stone episodes, prior medical or surgical treatment of nephrolithiasis, and young age of first stone event are all important components of the patient's history. | Structured and clearly defined referral criteria reflect thoughtful design that enhances appropriate implementation.   | Facilitator | Innovation    | Innovation Design     |

|                     |        |                                                                                                                                                                      |                                                                                                                                                                                                                |             |                        |                                  |
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|                     |        | Incomplete understanding, classification, and identification of VUS in a significant number of patients (53%)                                                        | VUS findings increase uncertainty and require advanced expertise, making implementation more cognitively and operationally challenging                                                                         | Barrier     | Innovation             | Innovation Complexity            |
|                     |        | In this model, we combine the knowledge and experience of health care workers that have particular interest and training in nephrolithiasis.                         | Combines individual expertise with strong internal collaboration, which supports coordinated implementation and shared learning.                                                                               | Facilitator | Inner Setting          | Relational Connections           |
|                     |        | The patient was also informed of the average cost of the genetic test, which would depend on their insurance status                                                  | Financial burden and uncertainty related to insurance can limit patient uptake of genetic testing.                                                                                                             | Barrier     | Outer Setting          | Financing                        |
|                     |        | Shared decision making without structured guidance contributed to inconsistent testing decisions.                                                                    | Engaging patients in decisions promotes uptake, trust, and alignment with individual values and needs                                                                                                          | Barrier     | Implementation Process | Engaging-Innovation Recipients   |
| Schott et al., 2025 | Canada | One of the cited barriers to accessing genetic testing for patients with CKD is the lack of knowledge among nephrologists on the genetic testing process             | This barrier reflects a gap in clinicians' understanding or confidence, which impacts their willingness or ability to engage with the innovation                                                               | Barrier     | Individual             | Innovation Deliverers-Capability |
|                     |        | Under-recognition and under-referral of patients with CKD for genetic testing.                                                                                       | This reflects a barrier stemming from clinicians' insufficient understanding or recognition of which CKD patients may benefit from genetic testing. It affects adoption at the individual level.               | Barrier     | Individual             | Innovation Deliverers-Capability |
|                     |        | Another cited implementation barrier for kidney genetics is the perceived lack of clinical utility following the establishment of a genetic diagnosis                | This reflects a belief barrier where clinicians may question the value or usefulness of the innovation (genetic diagnosis), especially if it doesn't clearly lead to actionable changes in care.               | Barrier     | Individual             | Innovation Deliverers-Motivation |
|                     |        | Clinical referral guidelines for kidney genetic assessment were initially developed through a review of the literature and were based on the best available evidence | This indicates that the innovation (i.e. referral guidelines for genetic assessment) is perceived as evidence-based and scientifically valid, which enhances stakeholder confidence and supports its adoption. | Facilitator | Innovation             | Evidence-base                    |

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|  |  | more selective criteria we employed to determine eligibility for genetic testing. We demonstrate that by instituting clinical referral criteria to a kidney genetic clinic, we can successfully identify a high proportion of patients with genetic kidney disease, with a short turnaround time for genetic testing results. |                                                                                                                                                                                                                                          |             |                        |                                          |
|  |  | By proceeding with a comprehensive testing strategy (i.e., comprehensive gene panel, exome sequencing and/or <i>MUC1</i> testing, if clinically indicated), we were able to confirm a genetic diagnosis in an additional 33 participants, thereby increasing the diagnostic yield from 23% to 34%                             | The innovation (comprehensive testing strategy) resulted in a tangible improvement in diagnostic yield, highlighting its superiority over more limited approaches—a clear relative advantage that supports implementation and uptake     | Facilitator | Innovation             | Relative advantage                       |
|  |  | All genetic results were reviewed by a multidisciplinary team                                                                                                                                                                                                                                                                 | The quote reflects the use of multidisciplinary collaboration and communication pathways within the organization to support accurate result interpretation and shared decision-making.                                                   | Facilitator | Inner Setting          | Relational Connections and communication |
|  |  | Ontario has a publicly funded health care system which covers the cost of gene panel testing for eligible patients                                                                                                                                                                                                            | This reflects a supportive external policy and funding environment that enables implementation by ensuring costs are not a barrier for eligible patients. Public funding is a critical facilitator of service uptake and sustainability. | Facilitator | Outer Setting          | Financing                                |
|  |  | We shared these guidelines with the nephrology, urology, pediatric, and genetic communities in our service area 3 months before establishing the clinic and then annually thereafter                                                                                                                                          | Actively communicating with relevant professional communities reflects deliberate engagement to foster collaboration and adoption.                                                                                                       | Facilitator | Implementation Process | Engaging-Innovation deliverers           |
|  |  | In whom variants of uncertain significance (VUS) were identified,                                                                                                                                                                                                                                                             | Routine reanalysis reflects deliberate efforts to monitor, evaluate, and adapt based on new                                                                                                                                              | Facilitator | Implementation Process | Reflecting and Evaluating                |

|                      |        |                                                                                                                                                                                                                                                                                  |                                                                                                                                                                                               |             |               |                                     |
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|                      |        | routine reanalysis was performed at an 18-month interval                                                                                                                                                                                                                         | information—key components of reflective practice in implementation.                                                                                                                          |             |               |                                     |
| Elliott et al., 2021 | Canada | The survey data showed patients are comfortable with the pathway and in the ability of nephrologists without specialized genetics training to provide pre-testing care                                                                                                           | Emphasizes patient-level comfort, trust, and acceptability of a new care model delivered by non-specialist providers                                                                          | Facilitator | Individual    | Innovation Recipients-Need          |
|                      |        | Use of commercially available gene panels allows any center with public funding to implement our mainstream pathway genetic testing without the need for in-house sequencing and interpretation capabilities                                                                     | The approach can be easily adapted and adopted by other centers, even those lacking internal genomic capabilities, which increases the feasibility and reach of the model.                    | Facilitator | Innovation    | Adaptability                        |
|                      |        | By eliminating the wait time associated with the initial clinic visit for assessment by medical genetics, the mainstream pathway returned results to patients faster and minimized the direct patient counseling by the genetics team for patients without an identified variant | The new pathway demonstrated a clear benefit over traditional workflows by improving turnaround time and resource efficiency, particularly by triaging counseling needs based on result type. | Facilitator | Innovation    | Relative Advantage                  |
|                      |        | We developed new resources for nephrologists to carry out pretest counseling and consent, selection of the appropriate testing panel, and arrangement for sample collection                                                                                                      | Reflects efforts to enhance access to information and practical tools that enable nephrologists to confidently and consistently engage in the genetic testing process.                        | Facilitator | Inner Setting | Access to Knowledge and Information |
|                      |        | Patient information handouts were developed to explain the testing, and genetic testing reports were developed for patients, and their health-care providers                                                                                                                     | These materials made it easier for clinicians and patients to access and understand genetic testing                                                                                           | Facilitator | Inner Setting | Access to Knowledge and Information |
|                      |        | We have successfully developed and implemented an efficient, mainstream approach to genetic testing in patients with ADPKD that leverages a collaboration between                                                                                                                | Because the success hinged on interdisciplinary collaboration, a key relational factor in implementation                                                                                      | Facilitator | Inner Setting | Relational Connections              |

|                         |        |                                                                                                                                                                                                                                                           |                                                                                                                                                                                                                   |             |                        |                                  |
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|                         |        | nephrology and medical genetics teams                                                                                                                                                                                                                     |                                                                                                                                                                                                                   |             |                        |                                  |
|                         |        | Expenses for testing were obtained through existing routine clinical coverage by Alberta's Genetic Resource Centre.                                                                                                                                       | This reflects an external policy and funding structure that facilitates implementation by covering the cost of genetic testing through existing clinical pathways—reducing financial burden and improving access. | Facilitator | Outer Setting          | Financing                        |
|                         |        | The group worked together over multiple sessions to adapt a standardized patient-centered approach from the oncology literature to work for patients with ADPKD. The pathway involved multiple iterations with input from all pathway design team members | The team modified a pre-existing model (from oncology) through multiple feedback cycles to suit a new context (ADPKD)                                                                                             | Facilitator | Implementation Process | Adapting                         |
| Pfirmann et al., 2021   | France | Patients had a large number of exams with a specific duration of exploration, in a limited time                                                                                                                                                           | Burdensome testing schedule within limited time frame                                                                                                                                                             | Barrier     | Innovation             | Innovation Complexity            |
|                         |        | The multidisciplinary evaluation in allowed us to obtain a follow- up of the patients and an evaluation of TSC targeting organs, according to the recommendations                                                                                         | Strong interprofessional collaboration enabling comprehensive, guideline-based evaluation and follow-up                                                                                                           | Facilitator | Inner Setting          | Relational Connections           |
| Becherucci et al., 2023 | Italy  | Thorough clinical (re-)evaluation of the patient by nephrologists specialized in genetic kidney disorders                                                                                                                                                 | Specialized nephrologists conducting thorough (re-)evaluation enhances diagnostic accuracy by applying expert knowledge in genetic kidney disorders                                                               | Facilitator | Individuals            | Innovation Deliverers-Capability |
|                         |        | Identifying criteria for referral of patients that avoided subjectivity and could increase the probability of selecting patients with genetic nephropathies                                                                                               | Objective referral criteria reduce subjectivity, improving the accuracy and consistency of selecting patients with genetic nephropathies, offering a clear advantage over less standardized approaches.           | Facilitator | Innovation             | Relative Advantage               |
|                         |        | A centralized straightforward clinical workup and whole exome sequencing analysis avoided the costs for multiple referrals and consultations                                                                                                              | A centralized, streamlined clinical workup and WES analysis offers clear advantages by reducing unnecessary referrals and consultations, improving efficiency and lowering costs.                                 | Facilitator | Innovation             | Relative Advantage               |

|                        |       |                                                                                                                                                                                                                            |                                                                                                                                                                                                                              |             |               |                                          |
|------------------------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------|---------------|------------------------------------------|
|                        |       | Organized procedure that centralized whole exome sequencing analysis in a single center to increase the diagnostic rate, a requirement for reaching cost-effectiveness and allocative efficiency.                          | Centralizing WES analysis in a single center optimizes the design of the diagnostic process, improving diagnostic rates and supporting cost-effectiveness and efficiency by utilizing specialized resources in one location. | Facilitator | Innovation    | Innovation Design                        |
|                        |       | The high costs of WES are considered a major hurdle for a broader implementation of genetic testing in clinical practice.                                                                                                  | The high cost of WES represents a financial burden directly tied to the innovation itself, impeding its broader implementation in clinical settings                                                                          | Barrier     | Innovation    | Innovation Cost                          |
|                        |       | Referring the patients to a single diagnostic center where WES equipment and multiple specialist expertise for reverse phenotyping are available, optimizing a procedure for patient re-evaluation and WES interpretation. | Referring patients to a single diagnostic center promotes strong collaboration between specialists, enhancing communication and teamwork in the re-evaluation and interpretation process, which improves patient care.       | Facilitator | Inner Setting | Relational Connections and Communication |
|                        |       | Integrated genetic and clinical data in multidisciplinary board discussions, including nephrologists with genetic expertise, pathologists, and nephrology-trained geneticists.                                             | Multidisciplinary board discussions integrating genetic and clinical data strengthen collaboration and knowledge sharing among specialists, supporting comprehensive and coordinated decision-making                         | Facilitator | Inner Setting | Relational Connections                   |
| Gutierrez et al., 2023 | Spain | Lack of genetic knowledge on the part of nephrologists.                                                                                                                                                                    | Reflects a gap in the knowledge, training, or expertise of nephrologists to understand or apply genetic information in clinical care                                                                                         | Barrier     | Individuals   | Innovation Deliverers-Capability         |
|                        |       | Lack of perceived benefit.                                                                                                                                                                                                 | Reflects low individual motivation or buy-in due to skepticism or uncertainty about the value or utility of genetic testing in clinical practice.                                                                            | Barrier     | Individuals   | Innovation Deliverers-Motivation         |
|                        |       | Difficulty in interpreting genetic variants                                                                                                                                                                                | Indicates the perceived challenge in understanding and applying variant classifications in clinical decision-making, adding to the cognitive load and implementation difficulty                                              | Barrier     | Innovation    | Innovation Complexity                    |
|                        |       | Need for pre- and post-test counseling                                                                                                                                                                                     | The need for specialized pre- and post-test counseling adds steps, time, and coordination to the clinical workflow, increasing the perceived complexity of implementing genetic testing.                                     | Barrier     | Innovation    | Innovation Complexity                    |

|                       |            |                                                                                                                                                                                                                                        |                                                                                                                                                                                                                                                 |             |               |                             |
|-----------------------|------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------|---------------|-----------------------------|
|                       |            | Presence of unexpected or unknown phenotypes such as allelic heterogeneity, incomplete penetrance, epigenetic regulation, mosaicism                                                                                                    | The biological complexities (e.g. allelic heterogeneity, incomplete penetrance, mosaicism) complicate variant interpretation and phenotype prediction, increasing the overall perceived difficulty of implementing genomic testing in practice. | Barrier     | Innovation    | Innovation Complexity       |
|                       |            | Creation of protocol for the early identification of genetic kidney disease, with all patients previously diagnosed in other nephrology outpatient clinics referred to this clinic.                                                    | Improves diagnostic accuracy and access to specialized care compared to usual outpatient management.                                                                                                                                            | Facilitator | Innovation    | Relative Advantage          |
|                       |            | Ability to perform a complete exome sequencing since 2019.                                                                                                                                                                             | Reflects access to a robust and validated diagnostic tool that enhances the clinic's ability to detect genetic kidney diseases.                                                                                                                 | Facilitator | Innovation    | Innovation Design           |
|                       |            | Support of the Genetic Service in the collaboration to perform additional evaluations, generating a genealogical tree for every patient who is going to undergo a specific genetic study due to suspicion of hereditary renal disease. | Reflects strong interdisciplinary collaboration that supports comprehensive assessment and coordinated care.                                                                                                                                    | Facilitator | Inner Setting | Relational Connections      |
|                       |            | Preparation of information sheet for the 18 primary care centers for the early identification of patients with cystic kidney disease or hereditary kidney disease.                                                                     | Builds formal linkages with primary care centers to improve early detection and referral of patients with suspected genetic kidney disease.                                                                                                     | Facilitator | Outer Setting | Partnership and connections |
|                       |            | Concern about costs and generalization in clinical practice                                                                                                                                                                            | Reflects external financial constraints and concerns about reimbursement or affordability of implementing genetic testing into standard practice.                                                                                               | Barrier     | Outer Setting | Financing                   |
| Caliment et al., 2024 | Luxembourg | Patients proposed for genetic testing were selected in accordance with the current international guidelines                                                                                                                            | Selection of patients based on international guidelines reflects reliance on high-quality, externally validated evidence, supporting the perceived strength and credibility of the innovation.                                                  | Facilitator | Innovation    | Innovation Evidence-base    |
|                       |            | Tiered-based genomic testing approach: Sanger sequencing when                                                                                                                                                                          | The structured, tiered design reflects thoughtful planning and optimization of the genomic                                                                                                                                                      | Facilitator | Innovation    | Innovation Design           |

|                           |        |                                                                                                                                                            |                                                                                                                                                                                            |             |               |                                  |
|---------------------------|--------|------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------|---------------|----------------------------------|
|                           |        | the index case was characterized. Otherwise, phenotype-based targeted panel testing followed by WES as a second line of testing                            | testing workflow to match clinical need and resource use                                                                                                                                   |             |               |                                  |
|                           |        | Patients with more complex presentations were offered a multidisciplinary consultation before an oriented genetic testing was proposed                     | Multidisciplinary consultation indicates effective teamwork and collaboration within the organization to support clinical decision-making.                                                 | Facilitator | Inner Setting | Relational Connections           |
| Pode-Shakked et al., 2022 | Israel | Team of experts well familiar with the genetic landscape of these disorders                                                                                | Highlights the clinical and genomic expertise within the team, which is a critical enabler for effective implementation of genetic services in nephrology.                                 | Facilitator | Individuals   | Innovation Deliverers-Capability |
|                           |        | This was very much due to dedication of the multidisciplinary team to pursue timely and efficient genetic analysis                                         | This highlights the intrinsic motivation and collective effort of the implementation team to deliver timely, efficient care.                                                               | Facilitator | Individuals   | Innovation Deliverers-Motivation |
|                           |        | Collaborative and dedicated service, in which pediatric nephrologists and medical geneticists evaluate the patient together, would be of synergistic value | This reflects a strong interprofessional collaboration model within the organization, enabling synergistic care and improved outcomes through real-time joint assessments.                 | Facilitator | Inner Setting | Relational Connections           |
|                           |        | Whenever possible, in the vast majority, with no costs for the patient and their family                                                                    | System-level financial support (e.g., public funding, institutional coverage, or insurance), which makes implementation feasible and acceptable to patients by removing cost as a barrier. | Facilitator | Outer Setting | Financing                        |

*MUC 1 = mucin 1, cell surface associated; MUC1fs = mucin 1 frameshift; VNTR = variable number tandem repeat; ADPKD = autosomal dominant polycystic kidney disease; PKD1 = polycystic kidney disease 1; PKD2 = polycystic kidney disease 2; ACMG = American College of Medical Genetics and Genomics; VUS = variant of uncertain significance; CAKUT = congenital anomalies of the kidney and urinary tract; CKD = chronic kidney disease; TSC = tuberous sclerosis complex; WES = whole exome sequencing*

## **Supplementary Figure Legends**

### **Supplementary Figure S1. Consolidated Framework for Implementation Research (CFIR) domains adapted for kidney genetics services**

This figure outlines the five CFIR domains contextualised for kidney genetics implementation: Outer setting (external context/policies/stakeholders outside the hospital), Inner setting (the hospital/health system where the kidney genetics clinic is embedded), Individuals (clinicians/patients/administrators directly interacting with kidney genetics clinics), Innovation (kidney genetics clinics/services), and Implementation process (key steps/strategies taken to plan, implement, and sustain kidney genetics clinics/services implementation). Adapted from The Centre for Implementation (2022).

### **Supplementary Figure S2. Matching of top implementation barriers to ERIC strategies using the CFIR-ERIC tool**

The six most frequently reported implementation barriers, prioritised by CFIR domain and construct, were systematically matched to implementation strategies using the ERIC taxonomy and CFIR–ERIC matching tool. For each barrier, ERIC strategies endorsed through expert consensus were extracted from the full set of 73 strategies. Endorsement percentages for each strategy–barrier pairing were summed across the prioritised barriers to generate cumulative percentage scores, reflecting the consistency of expert endorsement across multiple co-occurring determinants rather than relevance to a single barrier. Strategies are ranked by cumulative percentage, with higher values indicating broader cross-determinant applicability. Heatmap shading illustrates the relative contribution of individual CFIR domains to each strategy’s cumulative score. Abbreviations: CFIR = Consolidated Framework for Implementation Research; ERIC = Expert Recommendations for Implementing Change

### **Supplementary Figure S3. Implementation strategies proposed by global kidney genetics clinic leads**

Illustration of key implementation strategies suggested by survey respondents, grouped by ERIC domains. Commonly recommended strategies included stakeholder training and education, funding reform, workflow optimisation through formal implementation blueprints, and strengthened leadership engagement. Quotes reflect representative suggestions to improve integration, access, and sustainability of kidney genetics services.

Abbreviations: ERIC = Expert Recommendations for Implementing Change; ASN = American Society of Nephrology; NKF = National Kidney Foundation; ISN = International Society of Nephrology; FTE = full-time equivalent; MDT = multidisciplinary team; AI = artificial intelligence; WGS = whole genome sequencing; IT = information technology; EMR = electronic medical record.