

Kidney Genetics Implementation Scoping Review of Determinants Strategies and an Evidence Informed Global Roadmap

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Abstract

Introduction

Kidney genetics services are expanding globally, but implementation remains fragmented, variably resourced, and inequitable. We aimed to identify multilevel determinants of kidney genetics service implementation and align them with evidence-based strategies to inform a global implementation roadmap.

Methods

We conducted a scoping review following JBI methodology and PRISMA-ScR reporting guidelines, searching MEDLINE, Scopus, and Embase for English-language publications from 2000–2025. Determinants were coded using the Consolidated Framework for Implementation Research (CFIR) across innovation, individuals, inner setting, outer setting, and process domains. Priority barriers were matched to Expert Recommendations for Implementing Change (ERIC) strategies. Findings were triangulated with an international survey of kidney genetics clinic leads.

Results

Among 60 included studies, 28 reported implementation determinants. Barriers varied by region but most commonly included limited implementation team capability (n = 22), financing constraints (n = 16), service complexity (n = 14), policy and legal barriers (n = 10), resources limitations (n = 9), and low individual motivation (n = 8). Facilitators converged around innovation and inner setting domains, including perceived relative advantage (n = 12), design quality (n = 12), adaptability (n = 7), strong relational connections (n = 14), available resources (n = 8), and access to knowledge (n = 3). Survey responses from 48 international clinic leads corroborated these patterns. ERIC-matched priorities included utilizing financial strategies, developing stakeholder relationships, training stakeholders, and using evaluative and interactive strategies.

Conclusion

This CFIR–ERIC synthesis provides a theory-informed, strategy-linked roadmap for strengthening kidney genetics services and advancing equitable implementation globally.

Introduction

Chronic kidney disease (CKD) affects over 850 million people globally,¹ accounting for approximately 9% of the world's population.² It is among the fastest-growing causes of mortality worldwide, projected to become the fifth leading cause of death by 2050, with anticipated increases of 33% in age-standardized death rates and 28% in disability-adjusted life years if no urgent action is taken.^{3–5} In May 2025, the World Health Organization (WHO) recognized CKD as a global public health priority, calling for stronger systems for prevention, early detection, an equitable access to care.⁵

As genetic testing becomes increasingly integrated into nephrology practice, a growing proportion of CKD, across both pediatric and adult populations, can now be attributed to genetic etiologies.^{6–8} Despite growing evidence of its clinical and health system utility, the implementation of kidney genomics remains fragmented and inequitable across global settings, particularly in low- and middle-income countries (LMICs).^{9,10} Crucially, there is no consolidated global

evidence or policy roadmap addressing the key determinants, implementation strategies, and system-level actions needed to ensure equitable access to kidney genomics care. The absence of such guidance limits the translation of genomic advancements into routine nephrology services and perpetuates global disparities in diagnosis and care.

To address this gap, we undertook a two-part, mixed methods systematic investigation consisting of:

1. A scoping review of published literature on kidney genetics care, and
2. An international stakeholder consultation of global kidney genetics leads to identify real-world service barriers and enablers.

Determinants of effective implementation were first mapped onto the Consolidated Framework for Implementation Research (CFIR).¹¹ These determinants were subsequently linked to tailored implementation strategies from the Expert Recommendations for Implementing Change (ERIC) compilation,¹² and further aligned with WHO Health System Building Blocks¹³ and the Organisation's recent accelerating access to genomics for global health goals.¹⁴

Our ultimate objective is to offer a theory-informed, system-based, and policy-relevant roadmap to guide national and global stakeholders in advancing equitable kidney genetics care.

Methods

This study reports the distinct implementation-focused analysis from a prospectively designed scoping research program examining the global landscape of kidney genetics services comprised two complementary components: a scoping review of the published literature and an international stakeholder consultation. The previous published companion study characterized global kidney genetics clinic models, reported service, patient-reported, and implementation outcomes.¹⁵ In contrast, the present study synthesized implementation determinants, including barriers and facilitators, mapped corresponding implementation strategies, integrated stakeholder perspectives, and developed an evidence-informed global implementation roadmap. These analyses were not reported in the companion study.

The complete study protocol—including the eligibility criteria, search sources, search strategy, and study selection process was prospectively registered on the Open Science Framework (OSF; doi.org/10.17605/OSF.IO/N32VA). Methods specific to the present analysis are summarized below, shared methodological details and study instruments were reported in the companion study.¹⁵

Data sources

Scoping review

A scoping review was conducted in accordance with the Joanna Briggs Institute (JBI) methodology for scoping reviews¹⁶ and reported in line with the Preferred Reporting items for Systematic reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) reporting guidelines (**Supplementary Table S1**).¹⁷ Published literature reporting kidney genetics services was systematically identified and synthesized to elicit study characteristics, healthcare settings, determinants of kidney genetics service implementation, and implementation frameworks and theories used, if any.

International stakeholder consultation

An international stakeholder consultation was conducted using electronic survey of global kidney genetics clinic leads to contextualize scoping review findings and capture real-world implementation perspectives on determinants, as well as prioritized strategies to enhance the delivery of kidney genetics care. Participation was voluntary with implied consent through survey completion.

Full survey methods are detailed in **Supplementary Appendix S1**.

Findings from the scoping review and international stakeholder consultation informed the identification of key evidence gaps and provided the empirical analytic foundation for subsequent theory-informed analysis.

Study selection and extraction of implementation constructs

From the full set of studies identified through the scoping review, a subset was identified for focused implementation analysis. Studies were eligible if they reported—either explicitly (stated barriers or facilitators) or implicitly (qualitative quotes, narrative summaries)—on factors affecting the implementation of kidney genetics services. Eligible studies could reflect the perspectives of diverse stakeholders, including clinicians, patients, caregivers, administrators, or policy-makers.

Implementation-related data were extracted using a structured form including:

1. Barriers and facilitators
2. Implementation frameworks or theories (if any) applied in the included studies

Thematic coding and mapping to the CFIR framework

Both direct and indirect reporting of implementation determinants were included in the analysis. For studies that explicitly reported barriers or facilitators, these were extracted as stated. For studies that reported determinants indirectly through qualitative quotes, narrative descriptions, or interpretive summaries, relevant data were extracted and thematically coded as barriers or facilitators by two independent reviewers (R.S.L. and N.S.). Discrepancies were resolved through discussion and consensus.

CFIR framework was selected to guide determinant mapping because its comprehensive, multilevel structure supports the identification of barriers and facilitators across health-system, organizational, service-delivery, implementation process, clinician, and patient contexts.¹¹ Each barrier or facilitator was then deductively mapped to one of the five CFIR¹¹ domains, defined in this study as:

1. Innovation—The kidney genetics clinic and its core components, including the multidisciplinary care model and genomic testing approaches used in nephrology practice;
2. Outer setting—The external context outside the hospital or healthcare system shaping clinic implementation, including policies, funding and reimbursement, regulatory frameworks, patient needs, and broader stakeholder influences;
3. Inner setting—The hospital or healthcare system in which the kidney genetics clinic is embedded, including organizational structures, resources, leadership, and clinical workflows.
4. Individuals—Clinicians, genetic counselors, nurses, administrators, and patients and families who deliver, use, or are affected by the clinic; and
5. Implementation process—The strategies and actions undertaken to plan, implement, adapt, and sustain the clinic over time.

Further mapping aligned each data point to specific CFIR constructs within these domains. Definitions of the CFIR domains are summarized in **Supplementary Figure S1**.

Coding and thematic synthesis were conducted using a preliminary codebook developed in Microsoft Excel, which was iteratively refined during data extraction to ensure comprehensive coverage of implementation determinants across included studies. Frequency counts were performed to quantify the number and types of barriers and facilitators reported across CFIR domains and constructs, stratified by global region. Reporting frequency was used as a transparent and reproducible indicator of determinants recurring across heterogeneous health systems, as comparable data on broader prioritization criteria were inconsistently reported.

The final codebook—including study reference, country, extracted quotes, interpretation, assigned determinants, and mapped CFIR domain and construct—is presented in **Supplementary Table S2**.

Matching of barriers to implementation strategies using the ERIC taxonomy

Following thematic coding, the most frequently reported barriers—stratified by CFIR domain and construct—were identified based on frequency counts. These top-ranked barriers were then systematically matched to corresponding implementation strategies using the ERIC taxonomy,¹² selected for its standardized, consensus-derived taxonomy of discrete implementation strategies. The CFIR-ERIC matching tool, which is based on expert consensus on the suitability of ERIC strategies for specific CFIR determinants, was used to match prioritized barriers to implementation strategies. For each barrier, endorsed ERIC strategies (from the full set of 73) were extracted, and endorsement percentages were summed across co-occurring barriers rather than across barriers in isolation.¹⁸ Strategies were ranked by cumulative score, with the highest-ranking strategies prioritized as potentially high-impact interventions to address implementation challenges in kidney genetics service delivery.

Triangulation of findings from literature and stakeholder consultation

Common barriers and facilitators identified through stakeholder consultation were systematically coded using the CFIR framework and mapped to their respective constructs. In parallel, international stakeholders were asked to propose implementation strategies to address these barriers; these responses were coded and aligned with the ERIC taxonomy.¹² To enhance validity, we triangulated findings from the scoping review and the stakeholder consultation, prioritizing barriers and strategies that were consistently identified across both data sources. The final set of key candidate barriers and corresponding strategies was identified through convergence of published evidence and experiential data.

Alignment of recommended strategies with WHO health system building blocks and genomic access goals

The top ten implementation strategies were mapped to the six WHO health system building blocks¹³: health financing; access to essential medicines and technologies; leadership and governance; health information systems; health workforce; and service delivery. This alignment informed a proposed policy-relevant roadmap comprising actionable recommendations to enhance global access to kidney genetics services. The roadmap is guided by the WHO's strategic goals for accelerating genomic access¹⁹ namely, promotion of genomics through advocacy, implementation

of genomic methodologies, collaboration among entities engaged in genomics, and attention to the ethical, legal, and social issues (ELSI) raised by genomics.

Results

Study selection

The full study identification and selection process, including the PRISMA flow diagram, were described in the companion study.¹⁵ In total, sixty studies met the inclusion criteria. These were further assessed against predefined criteria for implementation-related content. Twenty-eight studies reported determinants relevant to the implementation of kidney genetics clinical services (Fig. 1), including four studies that reported implementation determinants explicitly, and twenty-four that reported such determinants implicitly through qualitative narratives, interpretations, or quotations. Among these, three studies applied a theory-informed implementation framework to guide analyses. Full study characteristics are summarized in Table 1.

Table 1
Characteristics of studies included in the implementation-focused synthesis (n = 28 studies)

Study Author, Year	Country	Study Design	Study Domains	Healthcare Setting/ Study Population	Type of Determinant Reporting	Framework Used
Jayasinghe et al., 2024 36	AUS	Retrospective and prospective cohort	Genomic utility, Implementation methodology	National network of 20 multidisciplinary kidney genetics clinics	Explicit	CFIR
Best et al., 2023 ³⁷	AUS	Cross- sectional	Implementation process and methodology	Non-genetic physicians based in Victoria and Queensland (interview- based)	Explicit	Process mapping; TDF
Kneifati- Hayek et al., 2024 ³⁴	USA	Cross- sectional	Stakeholder experience	Nephrologists across diverse settings (urban, rural, suburb) and academic institutions	Explicit	CFIR
Fernandez et al., 2024 35	USA	Cross- sectional	Stakeholder experience	Practicing paediatric nephrologists across the USA	Explicit	None
Mallet et al., 2016 ³⁸	AUS	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics clinic (Brisbane)	Implicit	None
Jayasinghe et al., 2021 39	AUS	Prospective cohort	Kidney genetics clinic model; Implementation methodology	Four kidney genetics clinics at tertiary hospitals in Melbourne	Implicit	None
Alkanderi et al., 2017 45	UK	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics clinic (Newcastle)	Implicit	None
Robinson et al., 2019 46	UK	Retrospective cohort	Kidney genetics clinic model; Stakeholder experience	Single tertiary hospital-based kidney genetics clinic (Cambridge)	Implicit	None

Determinant reporting refers to whether implementation barriers or facilitators were explicitly stated or implicitly derived from qualitative or interpretive data. See Supplementary Table 5 for full quotes and CFIR-coded constructs.

AUS = Australia; UK = United Kingdom; USA = United States of America; CFIR = consolidated framework for Implementation Research; TDF = Theoretical Domains Framework

Study Author, Year	Country	Study Design	Study Domains	Healthcare Setting/ Study Population	Type of Determinant Reporting	Framework Used
Elhassan et al., 2022 ⁴⁷	Ireland	Prospective cohort	Kidney genetics clinic model	National kidney genetics referral centre (Dublin)	Implicit	None
Thomas et al., 2020 ²⁰	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics clinic (Iowa)	Implicit	None
Thomas et al., 2021 ²¹	USA	Retrospective cohort	Kidney genetics clinic model; Implementation methodology	Single tertiary hospital-based kidney transplant centre (Iowa)	Implicit	None
Lundquist et al., 2020 ²²	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics clinic (Massachusetts)	Implicit	None
Amlie-Wolf et al., 2021 ²³	USA	Retrospective cohort	Kidney genetics clinic model; Implementation methodology	Single tertiary children's hospital-based kidney genetics clinic (Delaware)	Implicit	None
Pinto e Vairo et al., 2021 ²⁴	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics clinic (Mayo, Minnesota)	Implicit	None
Bekheirnia et al., 2021 ²⁵	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary children's hospital-based kidney genetics clinic (Texas)	Implicit	None
Bogyo et al., 2022 ²⁶	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics clinic (Columbia)	Implicit	None
El Ters et al., 2023 ²⁷	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney transplant centre (Mayo, Minnesota)	Implicit	None
Tan et al., 2023 ²⁸	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics	Implicit	None
<i>Determinant reporting refers to whether implementation barriers or facilitators were explicitly stated or implicitly derived from qualitative or interpretive data. See Supplementary Table 5 for full quotes and CFIR-coded constructs.</i>						
<i>AUS = Australia; UK = United Kingdom; USA = United States of America; CFIR = consolidated framework for Implementation Research; TDF = Theoretical Domains Framework</i>						

Study Author, Year	Country	Study Design	Study Domains	Healthcare Setting/ Study Population	Type of Determinant Reporting	Framework Used
				clinic (Cleveland, Ohio)		
McFadden et al., 2024 ²⁹	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based hereditary kidney tumour clinic (Indiana)	Implicit	None
Zhang et al., 2024 ³⁰	USA	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based general nephrology clinic (Philadelphia)	Implicit	None
Payne et al., 2024 ³¹	USA	Retrospective cohort	Kidney genetics clinic model; Implementation methodology	Single tertiary hospital-based multidisciplinary kidney stone centre (Mayo, Arizona)	Implicit	None
Schott et al., 2025 ³²	Canada	Prospective cohort	Kidney genetics clinic model; Implementation methodology	Single tertiary hospital-based kidney genetics clinic (Ontario)	Implicit	None
Elliott et al., 2021 ³³	Canada	Prospective cohort	Kidney genetics clinic model; Stakeholder experience	Single tertiary hospital-based general nephrology clinic (Alberta)	Implicit	None
Pfirmann et al., 2021 ⁴⁰	France	Retrospective cohort	Kidney genetics clinic model; Stakeholder experience	Single tertiary hospital-based dedicated tuberous sclerosis complex clinic (Bordeaux)	Implicit	None
Becherucci et al., 2023 ⁴¹	Italy	Prospective cohort	Kidney genetics clinic model	Single tertiary children's hospital-based kidney genetics clinic (Florence)	Implicit	None
Gutierrez et al., 2023 ⁴²	Spain	Retrospective cohort	Kidney genetics clinic model	Single tertiary hospital-based kidney genetics clinic (Madrid)	Implicit	None
<i>Determinant reporting refers to whether implementation barriers or facilitators were explicitly stated or implicitly derived from qualitative or interpretive data. See Supplementary Table 5 for full quotes and CFIR-coded constructs.</i>						
<i>AUS = Australia; UK = United Kingdom; USA = United States of America; CFIR = consolidated framework for Implementation Research; TDF = Theoretical Domains Framework</i>						

Study Author, Year	Country	Study Design	Study Domains	Healthcare Setting/ Study Population	Type of Determinant Reporting	Framework Used
Caliment et al., 2024 43	Luxembourg	Retrospective cohort	Kidney genetics clinic model	Single tertiary children's hospital-based kidney genetics clinic	Implicit	None
Pode- Shakked et al., 2022 ⁴⁴	Israel	Retrospective cohort	Kidney genetics clinic model	Single tertiary children's hospital-based kidney genetics clinic	Implicit	None
<i>Determinant reporting refers to whether implementation barriers or facilitators were explicitly stated or implicitly derived from qualitative or interpretive data. See Supplementary Table 5 for full quotes and CFIR-coded constructs.</i>						
<i>AUS = Australia; UK = United Kingdom; USA = United States of America; CFIR = consolidated framework for Implementation Research; TDF = Theoretical Domains Framework</i>						

Survey analytic context and international stakeholder consultation

To complement the published literature identified in the scoping review, the analyses also draw on an international stakeholder survey comprising 48 completed responses from kidney genetics clinic leads across multiple global regions. Respondents represented a senior, multidisciplinary cohort with substantial experience in nephrogenetics service delivery. Detailed respondent characteristics and survey response figures were reported in the companion study.¹⁵ The following sections present the implementation determinants and matched strategies informed by this combined evidence base.

Global determinants of implementation

Barriers

Barriers to the implementation of kidney genetics services identified through the scoping review were coded using the CFIR and summarized in Fig. 2. In North America, most reported barriers ($n = 61$)^{20–35} clustered within the outer setting ($n = 25$), followed by individual ($n = 18$), innovation ($n = 9$), inner setting ($n = 8$), and implementation process ($n = 1$) domains. In Australia and New Zealand (AUS/NZ), 46 barriers were identified,^{36–39} predominantly within the individual ($n = 15$) and inner setting ($n = 15$) domains, with fewer in the outer setting ($n = 6$), innovation ($n = 5$), and implementation process ($n = 5$) domains. In Europe, eight barriers were identified,^{40–44} mostly in the innovation ($n = 5$) and individual ($n = 2$) domains, with one in the outer setting and none in the inner setting or implementation process. In the United Kingdom (UK) and Ireland, where literature was limited, four barriers were identified^{45–47}, two each in the individual and innovation domains, reflecting regional variation in implementation challenges and reporting depth across global contexts.

To explore implementation challenges in greater depth, identified barriers were mapped to specific CFIR constructs within each domain and stratified by global region. The treemap in Fig. 3 visualizes the frequency and distribution of barriers across the five CFIR domains (individuals, inner setting, outer setting, innovation, and implementation process), with colour-coding by domain and proportional area by construct frequency.

In North America, barriers most frequently related to financing (n = 14) and policies and laws (n = 9) under the outer setting domain. Individual-level constructs included the implementation team's capability (n = 3) and motivation (n = 6), the capability and motivation of innovation recipients (n = 5), and the innovation deliverers' capability and motivation (n = 4). Innovation-related barriers reflected innovation complexity (n = 6), evidence-base (n = 2), and design (n = 1). Inner setting challenges related to available resources (n = 3), work and information technology infrastructure (n = 3), compatibility (n = 1), and access to knowledge and information (n = 1). A single barrier was coded to the implementation process, under engaging innovation recipients.

In AUS/NZ, the most frequently reported barrier was the capability of the implementation team (n = 12), followed by other individual-level constructs including capability of innovation deliverers (n = 2) and motivation of the implementation team (n = 1). Inner setting barriers included culture (n = 3), available resources (n = 4), access to knowledge and information (n = 2), work infrastructure (n = 2), structural characteristics (n = 1), relational connections (n = 2), and financing (n = 1). Outer setting barriers were coded to local attitudes (n = 3), policies and laws (n = 1), local conditions (n = 1), and financing (n = 1). Innovation-related challenges included complexity (n = 3), cost (n = 1), and design quality (n = 1). Implementation process barriers were distributed across doing (n = 3), engaging (n = 1), and reflecting and evaluating (n = 1).

In Europe and UK/Ireland, where data were limited, barriers were predominantly mapped to innovation complexity (n = 5), followed by individual-level capability of innovation deliverers (n = 2). Inner setting challenges included available resources (n = 2). Only one outer setting barrier was identified (financing), and none reported within the implementation process domain.

Key barriers to the implementation of kidney genomics were identified through surveys completed by 48 global kidney genetics clinic leads and categorized using the CFIR framework (Fig. 4). Unless otherwise stated, all percentages refer to survey respondents.

Barriers were most frequently reported within the individual domain, particularly regarding the implementation team's capabilities. More than 80% of survey respondents identified substantial knowledge gaps among nephrologists, including limited awareness of genetic kidney diseases, inadequate understanding of the utility of genomic testing, and a lack of formal genomic education or training. More than half cited uncertainty in independently ordering genetic tests, while 43% reported inappropriate referrals despite limited phenotype-genotype correlation.

Additional individual-level barriers included motivation of the implementation team, with 52% citing challenges keeping pace with rapidly evolving genomic knowledge, and capability of innovation recipients, such as low patient awareness and engagement (35%). Capability of innovation deliverers was also noted, with over a quarter of respondents reported discomfort communicating genetic results to patients.

Within the innovation domain, complexity was a major challenge, including limited access to functional studies and advanced testing platforms (70%), difficulties interpreting variants of uncertain significance (63%), limited local testing capability (59%), and administrative burden (50%). A further 69% highlighted insufficient patient-reported outcome measures as a key evidence gap.

Inner-setting barriers focused on available resources, with universal concern regarding shortages of genetic counselors and clinical geneticists (100%), sustainability challenges due to increasing demand (41%), weak relational connections among professionals (26%), and role ambiguity within multidisciplinary teams (21%).

Outer setting barriers included funding limitations related to restrictive national or institutional policies (40%), limited insurance coverage or high out-of-pocket costs (20%), and underrepresentation of patients in global population

genomic databases, limiting equitable and accurate variant interpretation (49%).

Facilitators

In parallel with the barriers, facilitators to kidney genetics service implementation were identified from the scoping review, coded to CFIR domains, and stratified by global region (Fig. 5). In North America, facilitators were most frequently mapped to the inner setting (n = 21), followed by innovation (n = 19), the implementation process (n = 7), the outer setting (n = 5), and the individual domain (n = 4).^{20–35} In Europe, 18 facilitators were identified,^{40–44} primarily within the innovation (n = 7) and inner setting (n = 6) domains, with fewer in the individual (n = 3), and outer setting (n = 2) domains. No facilitators were coded to implementation process domain.

In UK and Ireland, nine facilitators were identified,^{45–47} distributed across innovation, inner setting, and individual domains (n = 3 each), with none in the outer setting or implementation process.

In AUS/NZ, six facilitators were reported,^{36–39} predominantly within the innovation domain (n = 4), followed by individual, and implementation process (n = 1 each), with none reported in the inner or outer settings.

Aggregated facilitators were mapped to specific CFIR constructs to characterize key implementation enablers (Fig. 6). Most were coded to the inner setting, particularly relational connections (n = 14) and available resources (n = 8), followed by access to knowledge and information (n = 3), work infrastructure (n = 3), culture (recipient centredness) (n = 1), and communication (n = 1).

Within the innovation domain, facilitators most frequently reflected on relative advantage (n = 12), innovation design (n = 12), and adaptability (n = 7), with fewer references to evidence-based practice (n = 2). Implementation process facilitators included engaging innovation deliverers (n = 5), with single mentions of doing, reflecting and evaluating, and adapting.

Individual domain facilitators included capability of the implementation team (n = 3), innovation deliverers (n = 3), innovation recipients (n = 1), motivation (n = 2), and recipient need for genomics (n = 2). Outer setting domain facilitators included financing (n = 5), policies and laws (n = 1), and partnerships and connections (n = 1).

Survey respondents (n = 48) identified facilitators across all five CFIR domains (Fig. 7), with prominence in the inner setting, innovation, and individual domains.

Key individual domain facilitators included high capability and motivation among the implementation team, with respondents highlighting nephrologists with prior experience and training in nephrogenetics (90%) and team members experienced in genetic kidney diseases (46%). Strong motivation was also reported, as reflected by the multidisciplinary team's interest in nephrogenetics (63%) and active participation in genomics service delivery (56%).

The inner setting domain featured prominently, with enablers clustered across available resources, infrastructure, knowledge-sharing, relational connections, and communication. These included access to genetic counselors (69%) and their integration into service workflows (56%), administrative support (54%), education for multidisciplinary team (MDT) members (67%), inter-department collaborations (62%), synergistic nephrologist-genetics partnership (83%), and structured communication platforms such as multidisciplinary nephrogenetics boards (62%).

Innovation domain facilitators included evidence-based genomic service (70%), accurate phenotyping to guide pre-test evaluations (45%), highly adaptable clinic models with flexible workflows and specialist virtual consultations (62%), expedited results for urgent cases (43%), and access to advanced sequencing technologies (40%).

Implementation process domain facilitators emphasized regular feedback review (62%), whereas outer setting enablers included access to funding for genomic testing (60%), national kidney genomic networks (49%), and centralized collaboration platforms (40%).

Across regions, facilitators clustered predominantly within the innovation and inner setting domains, whereas barriers were frequently observed in the individual and outer setting domains. This pattern was similarly observed in survey responses from global kidney genetics clinic leaders.

Matched implementation strategies across regions

Using triangulated findings from the scoping review and international stakeholder survey, six high-frequency implementation barriers were identified:

1. Individual capability of the implementation team or innovation deliverers (n = 22),
2. Outer setting financing (n = 16),
3. Innovation complexity (n = 14),
4. Policy and legal barriers in the outer setting (n = 10),
5. Inner setting resource constraints (n = 9), and
6. Individual motivation (n = 8).

These were systematically mapped to evidence-based strategies for implementation using the ERIC taxonomy and CFIR-ERIC matching tool.^{12,48} Ten implementation strategies emerged across four ERIC domains, selected for their high cumulative percentage scores in addressing the above barriers (**Supplementary Figure S2**):

1. Use financial strategies including access new funding (177%), alter incentive/allowance structure (157%), and fund and contract for clinical innovation (107%).
2. Develop stakeholder interrelationships through identifying and preparing champions (149%), involving executive boards (123%), capturing and sharing local knowledge (112%), and informing local opinion leaders (93%).
3. Train and educate stakeholders by conducting educational meetings (105%)
4. Use evaluative and iterative strategies such as assess readiness and identify barriers and facilitators (97%) and develop a formal implementation blueprint (89%).

To further contextualize these theory-informed implementation strategies, free-text responses from international kidney genetics clinic leads were analyzed and mapped thematically to ERIC strategy categories (**Supplementary Figure S3**). Across ERIC strategy domains, substantial alignment was observed between strategies prioritized through CFIR-ERIC matching and those proposed by frontline implementers (Fig. 8). In particular, strategies related to utilising financial mechanisms, developing stakeholder relationships, and training and educating stakeholders demonstrated consistent convergence between high CFIR-ERIC prioritization and explicit practice-based endorsement.

Several strategies with strong theoretical support—such as accessing new funding, altering incentive or allowance structures, identifying and preparing clinical champions, involving executive boards, and capturing and sharing local knowledge were explicitly described by respondents as critical enablers for establishing and sustaining kidney genetics services. By contrast, strategies within evaluative and interactive domains, including assessing organizational readiness and identifying barriers and facilitators, showed weaker convergence, with limited practice-based articulation despite moderate theoretical prioritization.

Discussion

Through triangulation of a global scoping review and international stakeholder consultation, we identified six convergent CFIR determinants and ten matched implementation strategies that are both theory-informed and practice-grounded. This discussion synthesizes key implementation gaps, opportunities, and translates these findings into a systems-based, policy-relevant roadmap, the first to our knowledge, to advance equitable kidney genetics care across diverse healthcare system contexts.

Gaps and opportunities in global kidney genetics implementation

Lack of data and service development in low- and middle-income countries (LMICs)

Our review identified no published studies describing kidney genetics services in LMICs, highlighting a significant evidence and service-delivery gap across regions, including Africa, South America, and Asia. This reflects broader genomic inequities, as noted in a WHO consultation on community genetics, which identified persistent infrastructure limitations, workforce shortages, funding constraints, and low participation in global genomic research across LMICs.⁴⁹

Although Africa and Asia together account for the majority of the world's population, their genomic data remain underrepresented in global databases.¹⁰ This is particularly concerning given population-specific genetic risks—for example, individuals of African ancestry who are homozygous for *APOL1* risk alleles (G1/G2) face a two- to three-fold higher risk of CKD.^{50,51} Despite this, access to kidney genetics services remains limited in these regions.^{49,52}

Emerging initiatives, such as the H3Africa Kidney Disease Research Network (H3AKDRN)^{53,54} and national genomic program in Mexico⁵⁵ and India⁵⁶ are beginning to build capacity. However, widespread implementation of kidney genetics services in LMICs remains nascent.

Limited use of theory-informed, implementation science-guided approaches

Despite growing interest in integrating genomics into nephrology, few studies have employed formal implementation science methods to guide the design, evaluation, or scale-up of kidney genetics services. In our review, only 3 out of 28 studies (11%) explicitly used theory-informed frameworks to assess implementation determinants.^{34,36,37} This aligns with broader trends in genomics research, where only 1.7% of National Institutes of Health (NIH)-funded genomics grants include an implementation science component.^{57,58} The absence of structured frameworks increases the risk of misidentifying key barriers and applying generic, poorly tailored strategies that are less likely to address substantial barriers and thus hinder the implementation of novel genetic services. This contributes to ineffective resource use and implementation failure.

Importantly, our findings indicate that barriers to the implementation of kidney genomics are highly context-specific and vary by region. In North America, challenges predominantly arose from the outer setting, where access to genetic services remains heavily influenced by insurance coverage, pre-authorisation requirements, and fragmented funding structures. Despite the Genetic Information Nondiscrimination Act (GINA), enacted in 2008,⁵⁹ persistent concerns about insurability and employability continue to deter both clinicians and patients from engaging in genomic testing. By contrast, barriers in Australia and New Zealand were largely driven by inner setting factors. Respondents cited limited clinical genetics workforce capacity, insufficient institutional readiness, and an implementation climate that has not yet fully supported the integration of genomics into routine nephrology care.

Notably, across all regions, individual-level capability and motivation emerged as common barriers to implementation. Many clinicians reported low genomic literacy, uncertainty about clinical utility, and discomfort discussing genetic testing with patients. These individual and behavioural barriers compound the complexity of kidney genetics workflows—already characterised by lengthy turnaround times, multidisciplinary coordination, and perceived disruption to standard clinic operations—and together limit the scalability and sustainability of kidney genetics services globally.

A small but growing number of studies have begun to apply theory-informed frameworks, in collaboration with implementation scientists, to address these gaps. In Australia, for example, implementation science methods have been used to explore the attitudes and practices of nephrologists toward clinical genomics,⁶⁰ and to co-develop targeted education, training program, and strategies aimed at supporting the mainstreaming of renal genetic testing.^{61,62} These early efforts demonstrate the feasibility of embedding implementation frameworks into service development and capacity-building, and offer a valuable model for other regions aiming to accelerate the integration of genomics into routine kidney care.

Lack of patient and policy stakeholder involvement

A critical gap identified across studies is the absence of patient, family, and policy stakeholder voices in the implementation of kidney genetics services. Existing data on implementation determinants are overwhelmingly clinician-derived, with no studies involving patients, caregivers, or policy makers in identifying barriers or enablers. This provider-centric focus risks neglecting key factors related to patient experience, health equity, and system-level sustainability. Without meaningful engagement of care recipients and funders, kidney genetics services are likely to face challenges in uptake, scalability, and long-term integration into health systems. As genomic medicine advances, equitable and effective implementation will require the deliberate inclusion of patient and policymaker perspectives. Emerging global initiatives that centre patient and caregiver involvement in kidney disease research provide a valuable foundation.^{63–65} These models should be adopted and adapted by kidney genomic implementation efforts to ensure inclusive, context-responsive service design and policy translation.

Neglect of ELSI in kidney genetics care

None of the 28 studies included in our review evaluated the ELSI of implementing kidney genetics services. This represents a critical oversight, given that genetic testing raises complex issues that extend beyond the individual to affect families, communities, and health systems in ways few other medical technologies do.⁶⁶ A recent systematic review of ELSI in clinical genetics identified nine key domains—including directive counseling, psychosocial impact, disruption of family dynamics, stigma, and the influence of social determinants, religion, and culture on service acceptability—that warrant careful consideration.⁶⁷ In the absence of such inquiry, there is a real risk that kidney genomic services could inadvertently exacerbate existing inequities, particularly among marginalized populations and those in resource-limited settings.⁶⁶

Since the early 1990s, multiple global initiatives have sought to integrate ELSI research into genomic medicine, most notably the program established by the National Human Genome Research Institute (NHGRI) at the US National Institutes of Health.⁶⁸ These longstanding efforts offer a rich repository of frameworks and best practices that the global kidney genetics community can draw upon.⁶⁹ Moving forward, embedding ELSI evaluation into kidney genomics implementation research is essential to ensure equitable, ethically robust, and culturally sensitive service delivery.

From evidence to action: a policy roadmap for equitable kidney genetics care

Building on the critical gaps and barriers identified, this final section presents a global policy roadmap that systematically links these challenges to actionable strategies, health system levers, and WHO genomic goals to enable equitable access to kidney genetics care.

We propose a theory-informed, conceptual roadmap (Fig. 9) that integrates gaps, implementation determinants (CFIR), matched strategies (ERIC), WHO health system building blocks, actionable policy recommendations, and the WHO's four strategic genomic goals. This roadmap offers a practical framework to support countries and stakeholders in designing, implementing, and scaling equitable and sustainable kidney genetics services. It also highlights the urgent need to close the evidence-to-practice gap—without deliberate action, medical innovations may take over 17 years to reach routine patient care.⁷⁰ Embedding implementation science and health systems thinking is therefore essential to accelerate the translation of kidney genomics into real-world impact, ensuring timely access to accurate diagnosis, personalized therapies, and patient-centred care. Timely access to genomic medicine is not a luxury but a necessity for achieving equity in kidney health.

Strengths and limitations

This study, to our knowledge, provides the first comprehensive synthesis of global kidney genetics service implementation by triangulating findings from empirical literature and the lived experiences of kidney genetics clinic leads. Our theory-informed, system-level approach—grounded in implementation science frameworks (CFIR, ERIC)—enabled a robust mapping of real-world determinants to tailored strategies, health system levers, and WHO genomic goals. By integrating evidence from diverse sources and aligning them with global policy priorities, this work offers a practical and transferable roadmap to support countries in designing and scaling equitable kidney genetics services. The inclusion of both literature and primary survey data strengthens methodological rigour, while the use of implementation frameworks enhances theoretical depth. Importantly, this is the first study to explicitly link genomic implementation barriers to actionable policy strategies via the WHO health system building blocks, offering concrete guidance for translation into health policy and practice.

However, this study has limitations. Most included studies originated from high-income countries, limiting generalizability to LMICs with differing health system contexts. Reporting frequency of determinants reflects recurrence in the literature but not necessarily a determinant's importance, influence, addressability, timing or equity impact, and may underrepresent uncommon but consequential structural barriers. Some implementation determinants were inferred from narratives and may be subject to interpretation bias, despite independent coding and consensus. Survey responses were self-reported and thus prone to recall, non-response, and social desirability biases. Lastly, regional representation was uneven, with limited input from Asia, Africa, and Latin America, which may affect global applicability.

Conclusions

This review identifies key implementation gaps and barriers in delivering kidney genetics services and outlines candidate implementation strategies within an actionable policy roadmap aligned to WHO genomic goals. Advancing equitable kidney genomics will require sustained global collaboration to operationalize and adapt these strategies across diverse health systems. Future intervention studies should focus on implementing and rigorously evaluating these approaches to determine their impact on service delivery, patient outcomes, implementation effectiveness, and clarify the mechanisms through which change occurs.

Disclosure Statement

NS is the director of London Safety and Training Solutions Ltd, which offers training, improvement, and implementation solutions to healthcare organisations and the pharmaceutical industry. All other authors declare no relevant conflicts of interest.

Data Sharing Statement

This study includes data from publicly available sources and an international expert consultation of kidney genetics clinic leads. All data extracted from published studies are available within the article and supplementary materials. The survey data are not publicly available due to stakeholder confidentiality but may be shared in de-identified form upon reasonable request to the corresponding author, contingent upon institutional approval and a data use agreement.

Abbreviations

CFIR

Consolidated Framework for Implementation Research

Abbreviation: CFIR

Consolidated Framework for Implementation Research; AUS/NZ = Australia/New Zealand; UK = United Kingdom

Declarations

Competing Interests

NS is a Director of London Safety and Training Solutions Ltd, which provides training, improvement, and implementation solutions to healthcare organisations and the pharmaceutical industry. CQ is an Editor-in-Chief of npj Nephrology. In accordance with the journal's editorial policy, she will not participate in the editorial handling or decision-making for this manuscript. All other authors declare no competing interests.

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Author Contribution

RSL, NS, AJM, ECSY, CP, EB, and SA conceived and designed the study. RSL performed the data analysis and data visualisation and drafted the manuscript. CP, NS, and AJM critically revised the manuscript for important intellectual content. All authors reviewed and approved the final manuscript.

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Data Availability

This study includes data from publicly available sources and an international expert consultation of kidney genetics clinic leads. All data extracted from published studies are available within the article and supplementary materials. The survey data are not publicly available due to stakeholder confidentiality but may be shared in de-identified form upon reasonable request to the corresponding author, contingent upon institutional approval and a data use agreement.

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Appendix

List of global kidney genetics clinic leads collaborators (n = 48)

Full Name	Country	Affiliation
John Andrew Sayer	United Kingdom	Bioscience Institute, Newcastle University and The Newcastle upon Tyne Hospitals NHS Foundations Trust.
Richard Sandford	United Kingdom	University of Cambridge and Cambridge University Hospitals NHS Foundation Trust.
Jenny Patterson	United Kingdom	West of Scotland Regional Genomics Service.
Elizabeth Watson	United Kingdom	South West Genomics Laboratory Hub (NHS), Bristol.
Joanna Jarvis	United Kingdom	Department of Clinical Genetics, Birmingham Women's and Children's NHS Foundation Trust, Birmingham.
Helen M. Stuart	United Kingdom	Manchester Centre for Genomic Medicine, Manchester University NHS Foundation Trust, Manchester. Division of Evolution, Infection and Genomics, School of Biological Sciences, Faculty of Biology, Medicine and Health, University of Manchester, Manchester.
Anjali Menon	United Kingdom	North Bristol NHS Foundation Trust.
Daniel P. Gale	United Kingdom	UCL Centre for Kidney and Bladder Health, University College London and National Registry of Rare Kidney Diseases, Bristol, UK.
Melanie MY Chan	United Kingdom	Medical Research Council Laboratory of Medical Sciences, Imperial College London. Imperial College Healthcare NHS Trust, London.
Asheeta Gupta	United Kingdom	Department of Nephrology, Great Ormond Street Hospital, London. Department of Translational Sciences, University of Bristol, Bristol.
Peter J Conlon	Ireland	Beaumont Hospital, Dublin 9 and Royal College of Surgeons in Ireland.
Aron Chakera	Australia	Renal Unit, Sir Charles Gairdner Hospital, Nedlands.
Kathy Nicholls	Australia	The Royal Melbourne Hospital and the University of Melbourne, Parkville.
Randall James Faull	Australia	Renal Unit, Royal Adelaide Hospital, University of Adelaide.
John Whitlam	Australia	Department of Nephrology, Austin Health, Heidelberg, Victoria. Clinical Genetics Service, Austin Health, Heidelberg Victoria.
Michel Tchan	Australia	Dept of Genetic Medicine, Westmead Hospital, Sydney. Sydney Medical School, Sydney University, Sydney.
Hugh J McCarthy	Australia	Child and Adolescent Health, Faculty of Medicine and Health, University of Sydney.
Parthasarathy Shanmugasundaram	Australia	Department of Renal Medicine, St George hospital, Kogarah, New South Wales.

Full Name	Country	Affiliation
Catherine Quinlan	Australia	Department of Nephrology, Royal Children's Hospital. Kidney Regeneration, Murdoch Children's Research Institute. Department of Paediatric, University of Melbourne.
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Ella Wilkins	Australia	Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Flemington Road, Melbourne. Australia and Department of Paediatrics, University of Melbourne, Melbourne.
Andrew J. Mallett	Australia	Department of Renal Medicine, Townsville University Hospital, Townsville, Queensland.
Chirag Patel	Australia	Genetic Health Queensland, Royal Brisbane and Women's Hospital, Herston, Queensland.
Jessica Ryan	Australia	Monash Health and Monash University.
Kushani Jayasinghe	Australia	Department of Nephrology, Monash Medical Centre, Melbourne, Victoria. Monash University, Melbourne, Victoria. Department of Clinical genetics, Melbourne Health.
Amali Mallawaarachchi	Australia	Department of Clinical Genetics, Royal Prince Alfred Hospital, Sydney. Garvan Institute of Medical Research, Sydney.
Nicholas Cross	New Zealand	Department of Nephrology, Christchurch Hospital, Te Whatu Ora Waitaha Canterbury.
Ian Hayes	New Zealand	Genetic Health Service New Zealand-Northern Hub, Auckland.
Whitney Besse	United States	Department of Internal Medicine, Section of Nephrology, Yale School of Medicine, New Haven, Connecticut.
Xiangling Wang	United States	Departments of Kidney Medicine, and Medical Genetics and Genomics, Cleveland Clinic, Cleveland, Ohio.
Jordan G. Nestor	United States	Department of Medicine, Division of Nephrology, Columbia University, New York.
Amar J. Majmundar	United States	Department of Pediatrics, Boston Children's Hospital, Boston, Massachusetts.
Marie C Hogan	United States	Division of Nephrology and Hypertension, Mayo Clinic, Rochester, Minnesota.
Christie P Thomas	United States	Department of Internal Medicine, University of Iowa, Iowa City, Iowa.
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Figures

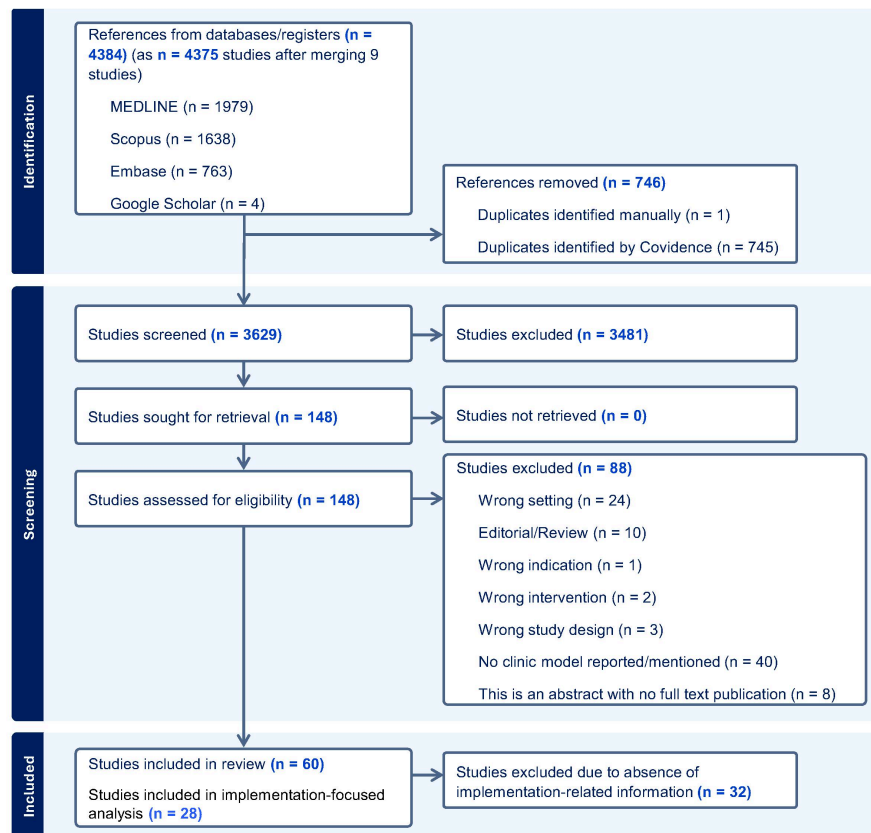


Figure 1

PRISMA flow diagram of study selection for implementation-focused analysis

A total of 4,375 records were identified through database searches: MEDLINE (n = 1,979), Scopus (n = 1,638), Embase (n = 763), and Google Scholar (n = 4). After removal of 746 duplicates, 3,629 records were screened by title and abstract, resulting in 148 full-text articles assessed for eligibility. Of these, 88 studies were excluded for reasons including wrong setting (n = 24), editorial/review articles (n = 10), wrong indication (n = 1), wrong intervention (n = 2), wrong study design (n = 3), no clinic model reported (n = 40), or abstract only with no full-text publication (n = 8). Of 60 studies identified in the scoping review of kidney genetics clinics, all were assessed for implementation-relevant content. 32 studies were excluded due to absence of implementation-related information. The remaining 28 studies were included in the implementation-focused analysis.

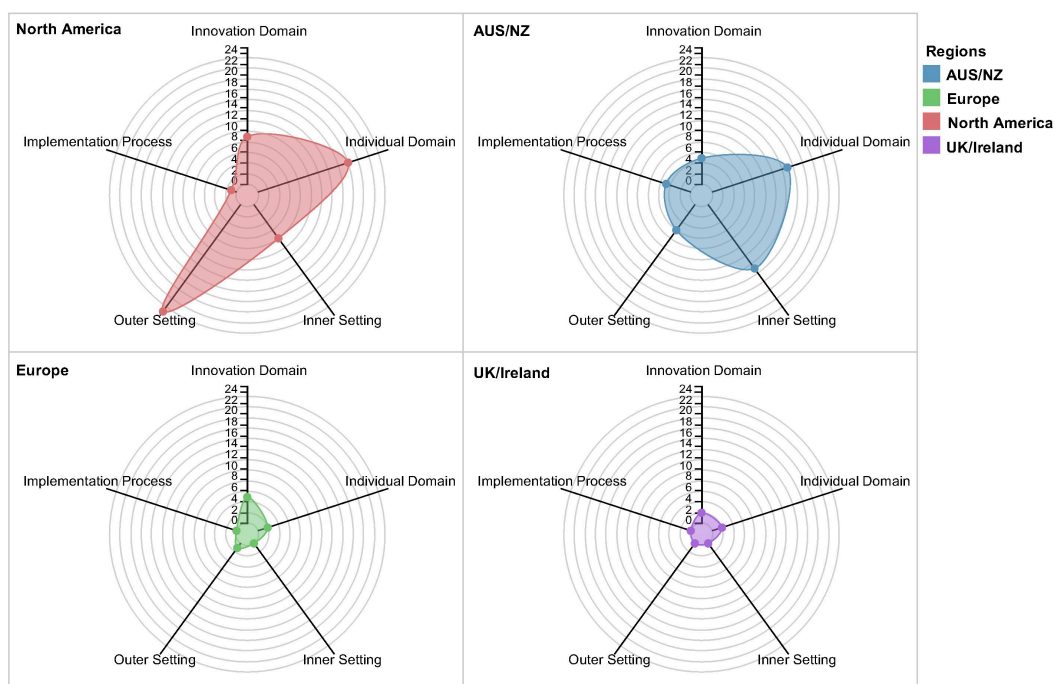


Figure 2

Distribution of implementation barriers across CFIR domains by global region

Radar plots showing the frequency of implementation barriers coded to each CFIR domain by region: North America, Australia/New Zealand (AUS/NZ), Europe, and United Kingdom (UK)/Ireland.

Abbreviation: CFIR = Consolidated Framework for Implementation Research.

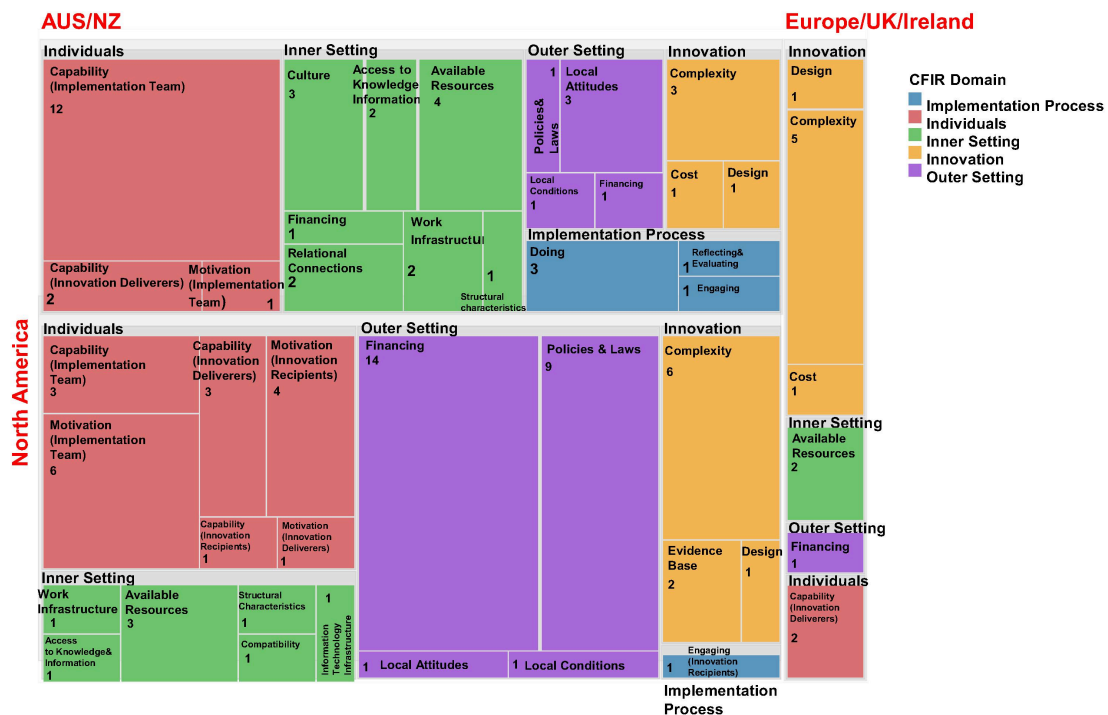


Figure 3

Combined treemap of implementation barriers by regions, CFIR domain, and construct

Treemap showing the frequency and distribution of reported barriers to kidney genetics service implementation across global regions, CFIR domains, and sub-constructs. Each coloured block represents a CFIR domain, subdivided into specific constructs (e.g., capability, financing, complexity), sized by the number of coded references.

Abbreviation: CFIR = Consolidated Framework for Implementation Research; AUS/NZ = Australia/New Zealand; UK = United Kingdom.

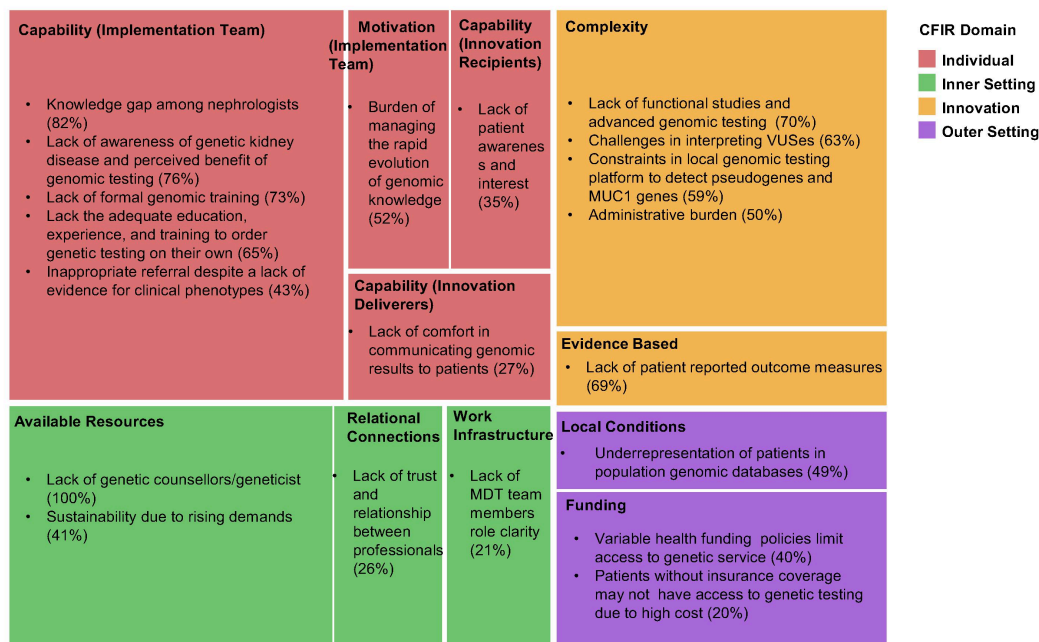


Figure 4

Survey-reported barriers to kidney genetics service implementation by CFIR domain and construct

Treemap summarising key barriers reported by 48 global kidney genetics clinic leads, organized by CFIR domains and constructs. Each coloured block represents a CFIR domain, subdivided into specific constructs, with block size proportional to the number of respondents reporting each barrier.

Percentages (%) indicate the proportion of respondents endorsing each barrier.

Abbreviation: CFIR = Consolidated Framework for Implementation Research; VUS = variant of uncertain significance; MDT = multidisciplinary team; *MUC1* = mucin 1, cell surface associated.

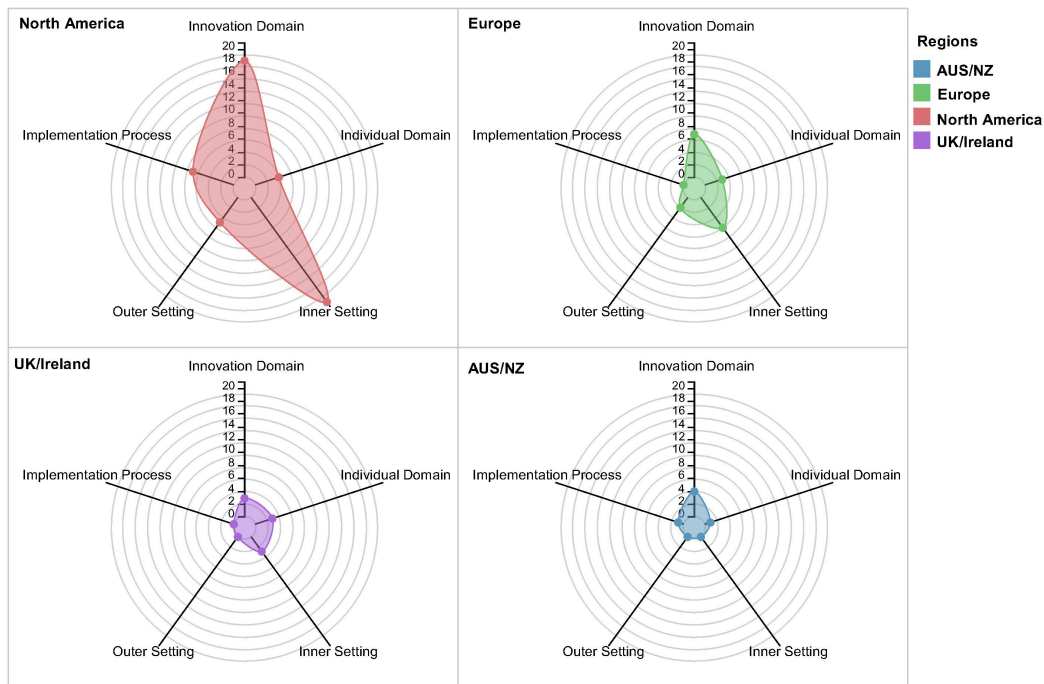


Figure 5

Distribution of implementation facilitators across CFIR domains by global region

Radar plots showing the frequency of implementation facilitators coded to each CFIR domain by region: North America, Australia/New Zealand (AUS/NZ), Europe, and United Kingdom (UK)/Ireland.

Abbreviation: CFIR = Consolidated Framework for Implementation Research.

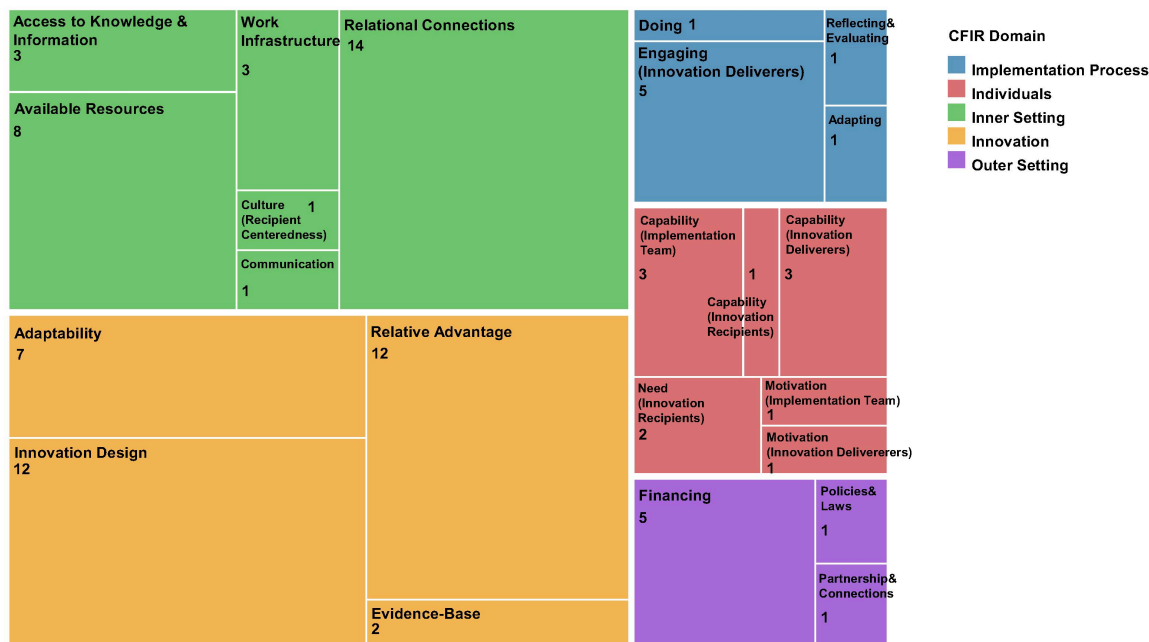


Figure 6

Combined treemap of implementation facilitators by CFIR domain and construct

Treemap showing the frequency and distribution of reported facilitators to kidney genetics service implementation across global regions, CFIR domains, and sub-constructs. Each coloured block represents a CFIR domain, subdivided into specific constructs, sized by the number of coded references.

Abbreviation: CFIR = Consolidated Framework for Implementation Research.

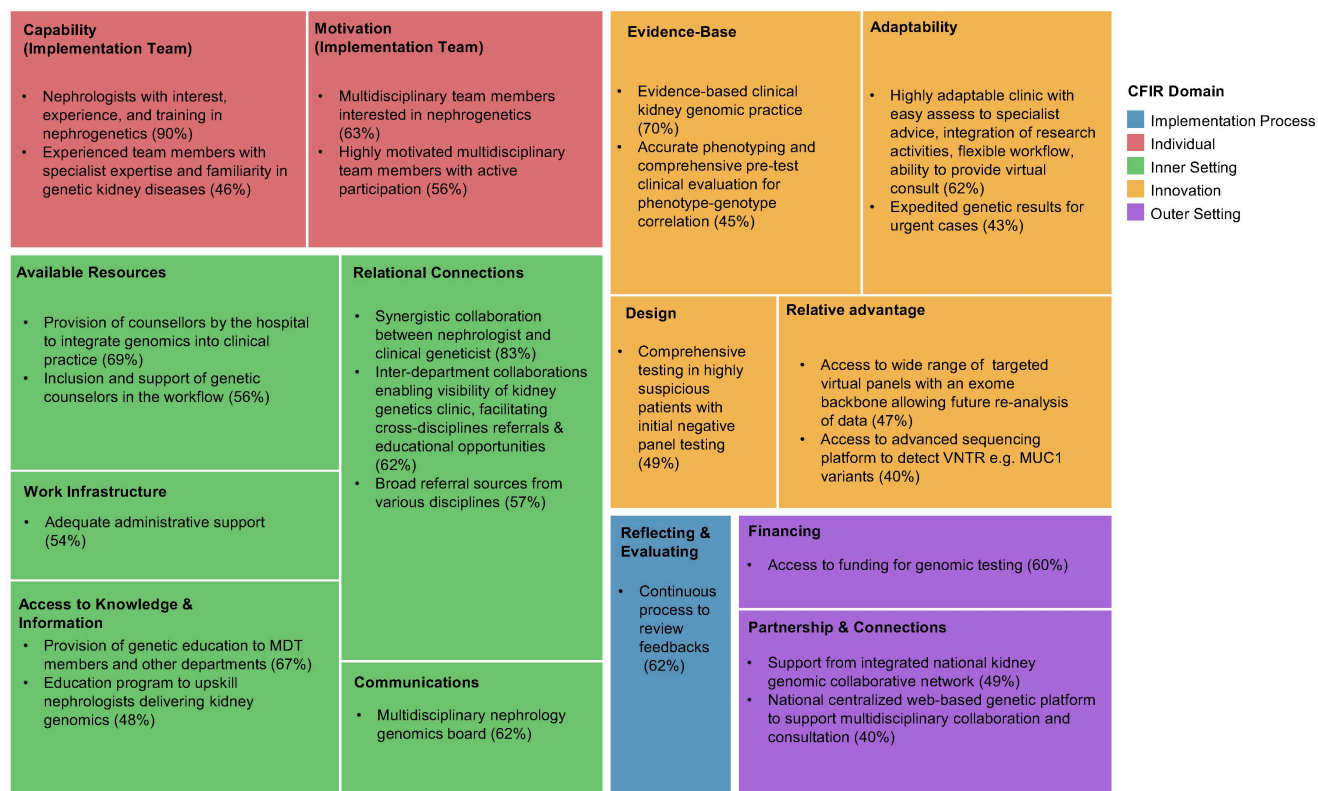


Figure 7

Survey-reported facilitators to kidney genetics service implementation by CFIR domain and construct

Treemap summarising key facilitators reported by 48 global kidney genetics clinic leads, organized by CFIR domains and constructs. Each coloured block represents a CFIR domain, subdivided into specific constructs, with block size proportional to the number of respondents reporting each facilitator.

Percentages (%) indicate the proportion of respondents endorsing each facilitator.

Abbreviation: CFIR = Consolidated Framework for Implementation Research; MDT = multidisciplinary team; VNTR = variable number tandem repeat, *MUC1* = mucin 1, cell surface associated.

ERIC Strategy Domain	Core Strategy	CFIR-ERIC Priority (Theory)	Survey Signal (Practice)	Triangulation
Utilize Financial Strategies	Access new funding	177%	●	✓
Utilize Financial Strategies	Alter incentive/allowance structure	157%	●	✓
Utilize Financial Strategies	Fund and contract for clinical innovation	107%	●	✓
Develop Stakeholder Relationships	Identify and prepare champions	149%	●	✓
Develop Stakeholder Relationships	Involve executive boards	123%	●	✓
Develop Stakeholder Relationships	Capture and share local knowledge	112%	●	✓
Develop Stakeholder Relationships	Inform local opinion leaders	93%	●	✓
Train and Educate Stakeholders	Conduct educational meetings	105%	●	✓
Use Evaluative and Interactive Strategies	Assess for readiness and identify barriers and facilitators	97%	○	—
Use Evaluative and Interactive Strategies	Develop a formal implementation blueprint	89%	●	✓

Figure 8

Triangulation of implementation strategies using CFIR-ERIC theory-based prioritisation and international survey signals

Implementation strategies are organized by ERIC strategy domains and core strategies. CFIR-ERIC priority reflects cumulative endorsement scores derived from expert-informed matching of prioritized CFIR determinants to ERIC strategies, with darker colour intensity indicating stronger theoretical support across multiple determinants (heat-map scale). Survey signal denotes qualitative evidence from international kidney genetics clinic leads, where ● indicates strategies explicitly proposed, ◻ implicitly described within broader responses, and ○ not reported. Triangulation symbols summarise alignment between theory and practice: ✓ indicates convergence between CFIR-ERIC prioritisation and survey data, △ indicates partial convergence, and — indicates theory-only prioritisation without corresponding practice signal. Survey strategies were analyzed qualitatively and mapped thematically; symbols reflect presence rather than frequency.

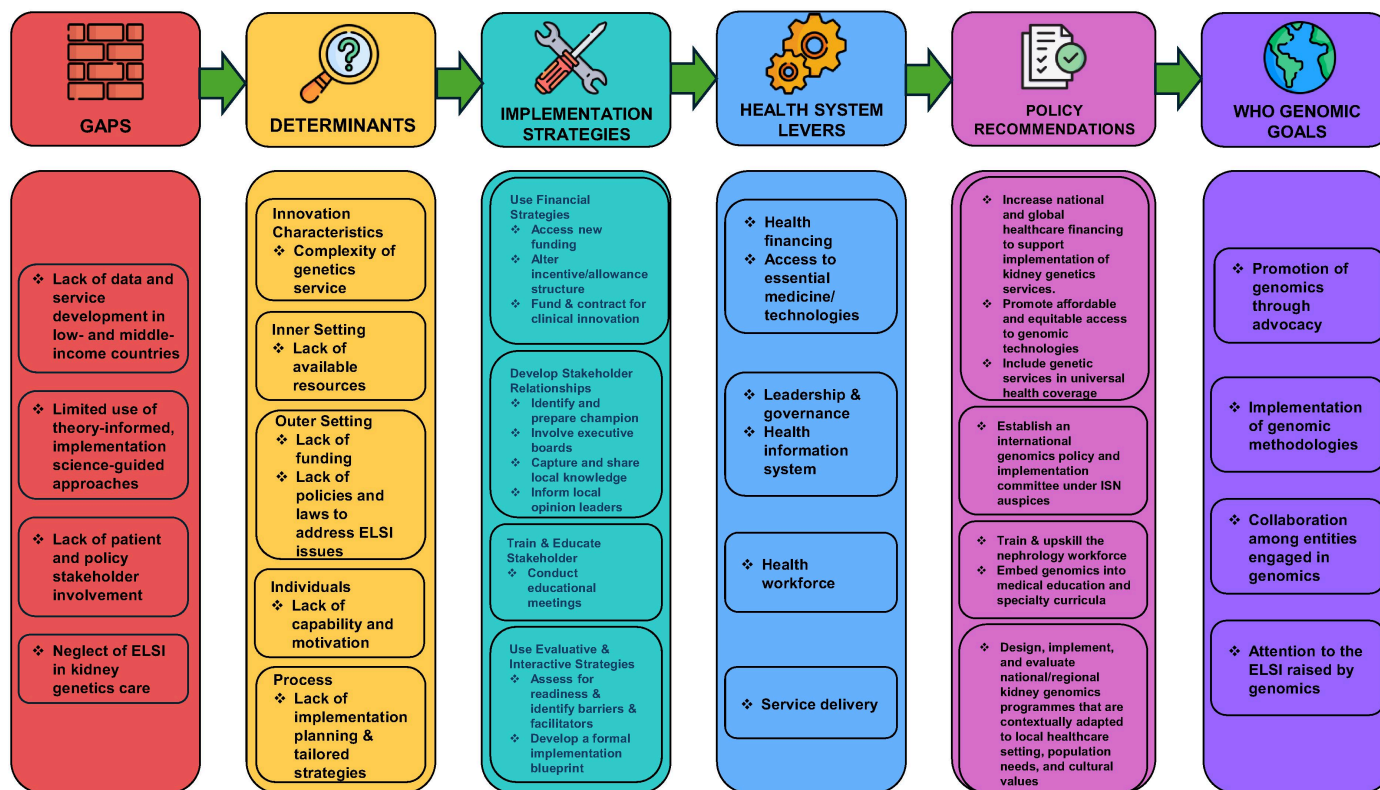


Figure 9

Conceptual policy roadmap to advance equitable kidney genomics implementation

A visual framework linking key implementation gaps to CFIR-based determinants, ERIC-aligned implementation strategies, WHO health system levers, actionable policy recommendations, and WHO's genomic goals. The roadmap highlights priority areas for global action including financing, stakeholder engagement, workforce capacity-building, and the design of contextually appropriate kidney genomics program to support sustainable and equitable kidney genetics service delivery.

Abbreviations: CFIR = Consolidated Framework for Implementation Research; ERIC = Expert Recommendations for Implementing Change; ELSI = ethical, legal, and social implications; WHO = World Health Organization; ISN = International Society of Nephrology.

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- [SupplementaryMaterial13Aug26.pdf](#)