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Assessing Physician Needs for the Implementation of Personalized Care

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Kidney diseases affect more than 690 million individuals worldwide and are associated with significant morbidity and mortality.[1] Hereditary nephropathies are genotypically and phenotypically heterogeneous and are often difficult to distinguish from "acquired" forms of kidney diseases (e.g., diabetes-associated nephropathy) because of overlapping, nonspecific features (e.g., elevated serum creatinine, proteinuria). Genome-wide sequencing techniques, such as exome and genome sequencing, are increasingly used in many clinical disciplines, including in nephrology. Recent studies show that genomic testing, like exome sequencing, can pinpoint the molecular etiology in 10% to 35% of cases of kidney disease.[2,3] Establishing a molecular diagnosis supports personalized management, such as guiding therapy, informing targeted workup and additional surveillance, and identifying at-risk family members for cascade screening.[4] Yet, despite the clinical utility of genomic testing, these diagnostic tools are not widely used in nephrology. A number of factors are likely to contribute to their limited clinical use, including the limited expertise of nephrologists in genomic medicine.[5]

Implementation of genomic testing in nephrology care relies, in large part, on providers' appreciation for a breadth of core knowledge specific to genomic medicine. This includes an understanding for specialized terminology, various diagnostic sequencing approaches (e.g., single-gene tests, gene panels, exome sequencing), categories of genomic results (e.g., polygenic risk scores, pharmacogenomic and genomic risk variants, primary diagnostic and otherwise medically actionable secondary findings, variants of uncertain significance, and other nondiagnostic results), and an awareness of complex ethical, legal, and technical considerations.[6] When nephrologists feel unsure about what test to order, how to interpret and discuss the findings with their patients, it can hinder their engagement in precision medicine efforts.[4] This is important because as genomic sequencing becomes more accessible, patients have growing opportunities to undergo testing, such as through their participation in genomic research, with expanded carrier screening as part of family planning, or through direct-to-consumer testing for ancestry.[7] Nephrologists will increasingly be faced with genomic data and called to interpret and apply these findings in the context of a patient's nephrology care. This issue is further complicated by the fact that there is a worldwide shortage of genomic professionals (e.g., genetic counselors, clinical geneticists) available to assist providers in the clinical implementation of genomic findings.[8,9] Therefore, systematic study of nephrologists' views and perceived self-efficacy implementing genomics into clinical nephrology practice are necessary for the advancement of the field.

In this issue of *Kidney International Reports*, Jayasinghe and collagues set out to assess Australian nephrologists' preparedness for the implementation of genomic medicine, within the context of their national health system. Using an anonymous electronic survey, which included items adapted from the IGNITE (Implementing Genomics in Practice) pre-implementation questionnaire and open-ended questions, the authors assessed attitudes and practices on genomic medicine and genomic testing among adult and pediatric nephrologists and nephrology trainees. Drawing from a convenience sample, the final analysis was performed on responses from 172 eligible participants, which the authors claim represents "at least 30% of practicing nephrologists and advanced trainees in Australia." The authors found most (77%) nephrologists had

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referred at least 1 patient for a genomic evaluation and that most (85%) believed genomic testing was clinically useful. Yet, a minority (23%) of respondents reported feeling confident applying the genomic results into clinical care. In addition, for more than half of respondents (57%), the preferred model of service delivery was referring patients to a multidisciplinary renal genetics clinic, which have been established throughout the country. Through qualitative and quantitative assessments, the authors also identified multiple perceived barriers nephrologists saw to using genomic testing as part of their diagnostic evaluations. These barriers included insufficient staffing, educational resources, and funding for referrals and testing.

Despite respondents' favorable views on the clinical utility of genomic testing, most felt unprepared to use genomic testing in clinical practice, and preferred referring patients to genomic professionals instead of ordering genomic testing and returning results themselves. These findings, along with nephrologists' perceived barriers to operationalizing routine use of genomic testing, provide valuable insights for the field of genomics and Precision Nephrology. First, the authors report that Australia has invested $87 million (USD) over the past 3 years on precision medicine programs, through state and national initiatives, and in collaboration with Australian Genomics and Melbourne Genomics Health Alliances, to promote integration of genomics across fields of medicine. In addition, with a national collaborative, *KidGen*, the authors report multidisciplinary *Renal Genetics Clinics* have been established throughout the continent to facilitate referrals and implementation. Yet, in spite of Australia's investment in infrastructure to support personalized care, and respondents enthusiasm for the availability of diagnostic sequencing approaches in nephrology, these findings suggest lack of familiarity in genomics (e.g., ordering testing, consent procedures, interpreting findings) still posed a significant barrier in their participation in these precision medicine programs. These findings underscore the need for further study into how providers' attitudes and perceived self-efficacy, influence utilization of genomic services. Next, the authors findings are a performance metric for Australia's personalized care initiatives, which may inform how future funding is allotted. In this study, most nephrologists supported the use of genomic testing. And, although it is unclear how Australia's precision medicine initiatives contributed to respondents' enthusiasm for integrating genomics into clinical practice, the findings suggest local and national investments in these programs successfully got providers' "buy-in". Therefore, instead of continued investments for programs that promote the clinical utility of genomic testing, the findings support the need for more research dedicated to addressing implementation challenges, such as providers' lack of preparedness using genomic data. Finally, this study highlights the importance of understanding the needs of providers, and the resources available to them, within the context of where they practice. Further studies into the specific informational and workflow needs of large cohorts of nephrologists, across diverse practice settings, are critical for the advancement of this field. For example, identifying barriers to using genomic testing that are specific to individual nations (e.g., third-party payer coverage for testing in the United States, funding for testing within Australia's health system), versus those potentially shared among nephrologists around the world (e.g., knowledge gaps in core clinical genomic concepts) will inform development of tailored solutions that can facilitate the integration of genomic information and guide patient care. Tools designed to address provider-specific needs, such as educational resources and clinical-decision support, can promote wider use of genomic resources and empower providers to use genomic data and deliver personalized care.

DISCLOSURE

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