

LETTER TO THE EDITOR

National survey on genetic test prescription in French adult nephrologists: a call for simplification and education

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Genomics has provided significant insight into the aetiology and mechanisms of kidney diseases in adult nephrology. Indeed, the recent use of exome or genome sequencing (GS) has opened new avenues for investigating adult nephropathies, particularly for those of unknown origins [1–5]. Between 10% and 48% of cases in this population could be attributed to a Mendelian disease. Since 2019, and in line with these advances, adult patients with chronic nephropathy of unknown aetiology with early onset (i.e. <45 years old) qualified for GS as part of the France Médecine Génomique 2025 plan, a French national genomics initiative. The plan has been set up as part of the healthcare system and is not considered a research program. Despite being free of charge for both the patient and the prescribing healthcare facility, as well as being promoted by academics, only a few adult nephrologists prescribe GS for their patients, with fewer than 80 genomes sequenced in this indication since the plan was initiated in 2019. Moreover, several works, such as the recent UK genomics project [6], have suggested that adult kidney disease management appears to be favourably impacted by genetic testing. In this context, we conducted a nationwide survey to investigate nephrologists' practices and, more broadly, their perception of genetic test prescriptions. The surveys were sent via several mailing lists (Club des Jeunes Néphrologues, Renaliste and oral communication at the last French Society of Nephrology meeting) and the link to the survey was presented at the 2021 French national nephrology congress.

Of the 1926 nephrologists registered in France in 2021 [7], 134 adult nephrologists from 66 different cities responded to

the survey (see Supplementary data for survey description and translation). The mean age of respondents was 40 years (interquartile interval 33; 44). The respondents were representative of different types of nephrological practice in France: practitioners from teaching hospitals (35%), general hospitals (26%), private not-for-profit hospitals (14%) and private clinics (26%).

The vast majority of nephrologists (75%) believe that genetic kidney diseases in adults are more common than it was thought 5–10 years ago, and yet only 12% of respondents regularly order genetic testing (i.e. once a month or more) (Figure 1A). The two main limiting factors were the complexity of the prescription (43%) and misunderstandings about which laboratories to send specimens to (47%) (Figure 1B). A total of 48% of nephrologists prescribing little or no genetic testing reported not referring patients they deemed eligible to a colleague or geneticist. A total of 63% of respondents were unaware of the existence of the Plan France Médecine Génomique 2025.

Only 14% of nephrologists felt adequately trained in the indications of prescription of a genetic test and 17% felt adequately trained in interpreting the results of a genetic report. Mirroring these results, 97% of respondents would be interested in training in prescribing and/or interpreting genetic results.

Finally, patient access to genetic tests was considered quite difficult with a median score of 29 (interquartile interval 20; 51), on a scale of 0 (very difficult) to 100 (very easy) (Figure 1C). Similarly, the turnaround times to gain genetic test results scored equally low at 30 (interquartile interval 18; 39), on a scale of 0 (much too long) to 100 (perfectly adequate)

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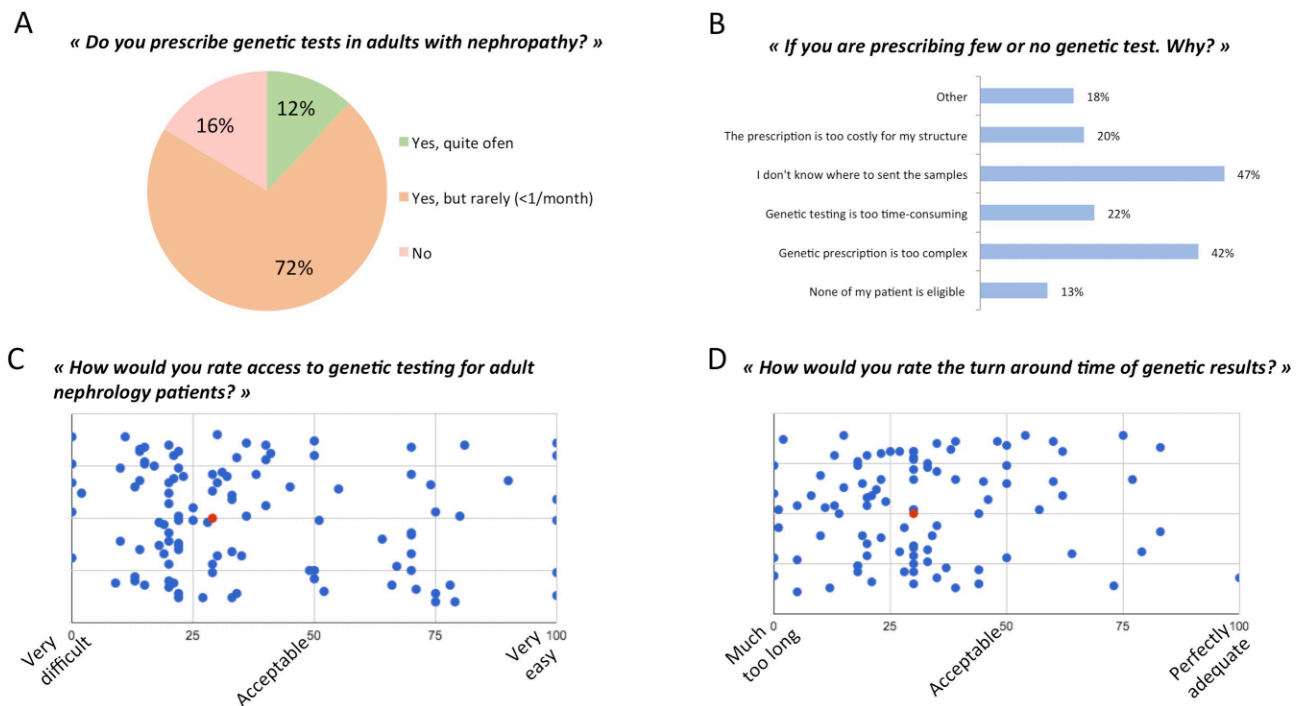


FIGURE 1: (A) Nephrologists' prescriptions habits. (B) Reasons for prescription limitations. (C) Nephrologists' perception of adult patients' genetic access. (D) Nephrologists' perception of the turnaround time for genetic results.

(Figure 1D). The optimal turnaround time was estimated as <2 months by 79% of nephrologists and from 2 to 6 months by 21% of nephrologists. No respondent answered 6–12 months.

To our knowledge, our nationwide survey among adult nephrologists is the first study to question clinicians, as potential prescribers of genetic tests, about their prescribing habits, their field experience and the obstacles to the prescription of genetic tests. The results of this survey highlight an important need for better information for, and outreach to, the nephrology community about the indications of genetic tests and the interpretation of results. Moreover, despite its important rare disease organization, Orkid (Orphan kidney disease network), and the existence of a national genomic plan accessible on behalf of their patients, adult nephrologists need adaptation of the current information on genomics. Respondents also expressed the need to simplify interactions between genetic laboratories and clinicians.

Although it is not exhaustive, our population of adult nephrologists appeared to be interestingly representative of the different modalities of practice and offered an overview of the whole country, with 66 cities represented. This survey was only conducted in France, which limits its general extrapolation, but adds to consideration of genomics as the turning point in nephrology in other countries. Our results are in line with a US survey based on self-evaluation of competency after a nephrology fellowship in which a significant percentage of respondents reported receiving insufficient or no training to feel competent in genetic renal disease [8]. A US national survey in post-graduate residents demonstrated significantly lower perceived understanding of genetics compared with non-genetic topics [9].

The rapidity with which genomics has become popularized over the past few years in adult nephrology means that ma-

ior challenges remain. In particular, despite being available free of charge, inequality of genetic test access for renal patients appears exacerbated by under-education of adult nephrologists. Giving nephrologists the appropriate tools to engage actively in the genetics care of their patients could accelerate access to genetic testing. Along with a recent publication revisiting the roles of primary care clinicians in genetic medicine [10], it is clear that clinicians are central in genomics, as they are the ones initiating genetic testing, capturing the patient's medical history, building the family pedigree and potentially helping with genetic variant co-segregation in the family. In order to ensure that nephrologists do not miss the boat of genomics, genomics should be integrated into the nephrology curriculum, with post-graduate education offered much more widely.

SUPPLEMENTARY DATA

Supplementary data are available at [ckj](https://doi.org/10.1111/ckj.14111) online.

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DATA AVAILABILITY STATEMENT

All data on which the conclusions of the article rely are available to readers.

CONFLICT OF INTEREST STATEMENT

None of the authors declares any competing interests. The authors declare that the results presented in this article have not been published previously in whole or in part.

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