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CLINICAL RESEARCH

Black Patients Equally Benefit From Renal Genetics Evaluation but Substantial Barriers in Access Exist

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Introduction: Genetic testing is increasingly accessible to patients with kidney diseases. Racial disparities in renal genetics evaluations have not been investigated.

Methods: A cohort of patients evaluated by the Cleveland Clinic Renal Genetics Clinic (RGC) from January 2019 to March 2022 was analyzed.

Results: Forty-eight Black patients, including 27 (56.3%) males, median age 34 (22–49) years and 232 White patients, including 76 (32.8%) males, median age 35 (21–53) years, were evaluated. Black patients were more likely to have end-stage kidney disease (ESKD) at the time of referral compared with White patients (23% vs. 7.3%, $P = 0.004$), more likely to be covered by Medicaid (46% vs. 15%, $P < 0.001$), and less likely to be covered by private insurance (35% vs. 66%, $P < 0.001$). Black patients were more likely to "no show" to scheduled appointment(s) or not submit specimens for genetic testing compared with White patients (24.1% vs. 6.7%, $P = 0.0005$). Genetic testing was completed in 35 Black patients. Of these, 37% had a positive result with 9 unique monogenic disorders and 1 chromosomal disorder diagnosed. Sixty-nine percent of Black patients with positive results received a new diagnosis or a change in diagnosis. Of these, 44% received a significant change in disease management. No differences in diagnostic yield and implications of management were noted between Black and White patients.

Conclusion: Black patients equally benefit from renal genetics evaluation, but barriers to access exist. Steps must be taken to ensure equitable and early access for all patients. Further studies investigating specific interventions to improve access are needed.

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Genetic kidney diseases are not uncommon. The global prevalence of chronic kidney disease (CKD) is estimated to be between 11.7% and 15.1%. Approximately 4.9 to 7.0 million of these patients are living with kidney failure requiring renal replacement therapy.[1-3] Exome sequencing (ES) in a combined cohort of more than 3000 patients with CKD yielded a genetic diagnosis in nearly 10% of cases.[4] In the last decade, genomic studies have facilitated significant advancements in the diagnosis and management of patients with kidney diseases. The discovery of the *APOL1* G1 and G2 risk alleles in individuals of recent African ancestry is one such example.[5] To date, approximately 625 genes have been significantly associated with kidney disease.[6-10]

Black individuals are almost 4 times as likely as White individuals to develop kidney failure. Although Black individuals make up approximately 13% of the population, they account for 35% of people with kidney failure in the United States.[11,12] CKD results from complex interactions between individual genetic and environmental factors. Disparities in kidney disease outcomes are largely driven by environmental factors, including access to healthy foods, neighborhood and

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housing quality, insurance, stress, access to healthcare, and more.[11,13] Genetic risk factors may also play a role. For example, *APOL1* nephropathies are driven by G1 and G2 risk alleles and have a high prevalence in the Black population (~13%).[14] Genetic assessments produce important diagnostic and prognostic information which facilitates individualized treatment plans and aids in family planning.[5] Despite the disease burden and renal risk variants, there is little information on the prevalence of genetic testing in Black individuals with kidney disease.

RGC of the Cleveland Clinic was founded in December of 2018 and has grown rapidly with more than 300 patients evaluated.[15] All genetic testing was performed in Clinical Laboratory Improvement Amendments-certified labs. In this study, we compared the clinical characteristics, diagnostic modalities, and diagnostic implications of Black and White patients with nephropathy referred to the RGC in the Cleveland Clinic.

METHODS

Study Population

This study was approved under IRB18-705 by the Cleveland Clinic Foundation Institutional Review Board. Patients consented and were enrolled from January 2019 through March 2022. Patients were evaluated by the RGC—a collaborative program between the Center for Personalized Genetic Healthcare and Glickman Urological and Kidney Institute. Review of medical records was performed independently by 2 researchers (CB and XT). Data collected includes clinical and laboratory characteristics, such as self-identified race, modalities of genetic evaluation, diagnostic yield of genetic testing and its implications of diagnosis and management, referring provider, and insurance coverage. Estimated glomerular filtration rate for all patients was calculated using the 2021 Chronic Kidney Disease Epidemiology Collaboration Creatinine Equation.[16] Data on "no shows" to scheduled RGC appointment(s) or failure to submit samples despite the decision to undergo genetic testing and RGC patient outreach were collected. Data on "no shows" to scheduled General Nephrology Clinic on the main campus of Cleveland Clinic from January 2019 through March 2022 was also analyzed. As part of standard care of the Cleveland Clinic, all patients received a confirmation message with detailed instructions for their upcoming visit via MyChart, text message, or, in rare cases, mail, immediately after their appointment has been scheduled, followed by a reminder message 1 to 2 weeks before the appointment and a second reminder message 1 to 3 days before the scheduled visit. The reminders were tailored to the patient's preferences, with email reminders sent out within 7 days of the appointment to remind patients to precheck-in. Paper reminders were sent out 2 weeks before the appointment, whereas phone reminders were sent 1 to 3 days before the appointment, unless the patient opted out. Text reminders were also available for patients who opted-in and were sent 1 to 2 days before the appointment.

RGC Evaluation

As described previously, the clinic visit entailed an interview with 1 genetic counselor to collect history, including 3 generations of family history, and a physician evaluation for genetic testing decision and disease management.[15] All patients received pretest counseling, including the review of the Genetic Information Nondiscrimination Act.[17,18] Genetic testing was not ordered until patient insurance preauthorization was obtained, or the patient agreed to pay testing expenses. Patients for whom insurance coverage was denied or who were unable to pay were offered sponsored testing if clinically appropriate. Sponsored testing is supported by commercial vendors at no cost to patients. Testing results were interpreted jointly by the ordering physician and the genetic counselor. Negative testing results or variants of unknown significance were reported to patients by the genetic counselor. Positive results were reviewed with the ordering physician and a follow-up visit was offered.

Genetic Testing

DNA samples were collected from buccal swab or blood. Genetic testing was performed in a Clinical Laboratory Improvement Amendments-certified laboratory. Laboratories utilized include GeneDx (Gaithersburg, MD), PreventionGenetics (Marshfield, WI), Natera (Austin, TX), Blueprint Genetics (Seattle, WA), Invitae (San Francisco, CA), Otolaryngology and Renal Research Laboratories of the University of Iowa (Iowa City, IA), Genetics and Genomics Diagnostic Laboratory at the Cincinnati Children's Hospital Medical Center, Cleveland Clinic Molecular Genetics Laboratory (Cleveland, OH), and Mayo Clinic Molecular Genetics Laboratory (Rochester, MN). Genetic testing modalities utilized include chromosomal microarray, single gene test, multigene panel, and ES. ES may include mitochondrial gene sequencing. The selection of genetic testing modalities and strategies was guided by clinical evaluation. When there was a strong suspicion of a particular disorder, a tiered testing approach was employed, beginning with a single gene test, and if the results were negative, followed by a reflex to a multigene panel. When the clinical differential diagnosis was

broad, a multigene panel was used as the first-tier test. ES is usually selected as a second-tier or third-tier test. Genetic testing results were analyzed and interpreted according to the American College of Medical Genetics guidelines.[19]

Genetic testing results were categorized into 5 groups as follows: (i) a positive result was defined as a pathogenic or likely pathogenic variant in an autosomal dominant or X-linked disorder, or homozygous or compound heterozygous pathogenic or likely pathogenic variants in an autosomal recessive condition; (ii) carrier of autosomal recessive conditions, including individuals with a heterozygous pathogenic or likely pathogenic variant in an autosomal recessive disorder; (iii) 2 APOL1 kidney disease risk alleles (G1 (rs73885319, p.S342G) and G2 (rs71785313, p.N388_Y389del)) in the homozygous or compound heterozygous state (G1/G1, G2/G2, or G1/G2); (iv) variant of unknown significance; and (v) negative result.

Statistical Analysis

Patient characteristics were summarized by frequency with proportion for categorical data and medians with interquartile ranges for continuous data. Chi-squared method, with addition of Monte Carlo simulation method when needed, was used to test for differences in categorical variables.[20,21] Unpaired 2-sample Wilcoxon tests were used to compare continuous variables. Odds ratio *P*-values were obtained by Fisher Exact testing. All statistical analyses were performed using R version 4.1.3 (The R Foundation for Statistical Computing, Vienna, Austria).

RESULTS

Characteristics of Study Sample

A total of 307 new patients from 299 pedigrees were evaluated at the RGC of Cleveland Clinic main campus between January 2019 and March 2022 and consented to this study. Of these, 48 patients self-identified as Black and 232 patients self-identified as White. The characteristics of Black and White patients in the RGC are listed in Table 1. Of the remaining 27 patients, 13 self-identified as multiracial or multicultural, 2 self-identified as multiracial or multicultural and Hispanic, 6 self-identified as Asian, and 6 either declined to provide race or racial data was unavailable.

There were significantly fewer female Black patients compared with female White patients (44% vs. 67%, $P = 0.003$). Black patients were more likely to be with ESKD at the time of referral to RGC compared with White patients (23% vs. 7.3 %, $P = 0.004$). Among the rest who were not with ESKD, median estimated glomerular filtration rate at index visit was not significantly different between Black and White patients (80.5 [47–100] ml/min per 1.73 m^2 vs. 90 [52–113] ml/min per 1.73 m^2) ($P = 0.20$). Fifty-two percent (25 of 48) of Black patients had a family history of kidney disorder, which did not differ from White patients. Glomerular disease is the leading reason Black patients presented to the RGC, followed by cystic kidney diseases and electrolytes disorders. No significant differences in clinical presentations or referral reasons were identified across racial groups. Black patients were significantly more likely to be covered by Medicaid compared with White patients (46% vs. 15%, $P < 0.001$). In contrast, fewer Black patients were covered by private insurance compared with White patients (35% vs. 66%, $P < 0.001$). Visits were conducted via distance health for 16.3% of Black patients and 24.6% of White patients ($P = 0.29$).

As shown in Table 2, the combined analysis showed that Black patients were more likely to "no show" to scheduled RGC appointment(s) or not submit samples despite the decision to undergo genetic testing and RGC patient outreach (24.1% vs. 6.7%, $P = 0.0005$). Specifically, Black patients were more likely to not show up to scheduled RGC appointment(s) compared with White patients (11.1% vs. 2.5%, $P = 0.006$).

Table 1. Characteristics ofBlack and White patients seen at index visit at the Renal Genetics Clinic of the Cleveland Clinic from January 2019 through March 2022

Factor	Black ($N = 48$)	White ($N = 232$)
Age, Median (IQR)	34 (22–49)	35 (21–53)
Males[a], No. (%)	27 (56.3)[a]	76 (32.8)
eGFR, ml/min per 1.73 m^2, median (IQR)	80.5 (47–100)	90 (52–113)
Family History, No. (%)	25 (52.0)	125 (53.9)
ESKD[a], No. (%)	11 (22.9)[a]	17 (7.3)
Dysmorphic features, No. (%)	7 (14.5)	18 (7.8)
Presentations, No. (%)		
• Glomerular disease	19 (39.6)	73 (31.5)
• Cystic kidney disease	12 (25)	59 (25.4)
• Electrolytes disorder	9 (18)	58 (25)
• CAKUT	2 (4.1)	13 (5.6)
• Kidney Stones and/or Nephrocalcinosis	0 (0)	10 (4.3)
• Renal vascular disease	1 (2)	2 (1)
• Tubulointerstitial disease	0 (0)	2 (1)
• Angiomyolipoma	0 (0)	1 (0)
• Unclear diagnosis	3 (6.3)	11 (4.3)
Insurance status, No. (%)		
• Medicaid[a]	22 (45.8)[a]	34 (14.7)
• Medicare	9 (18.8)	39 (16.8)
• Private[a]	18 (37.5)[a]	152 (65.5)
• Military[a]	0 (0)	5 (2.2)
• International	0 (0)	1 (0.4)

CAKUT, congenital anomalies of kidneys and urinary tract; eGFR, estimated glomerular filtration rate; ESKD, end-stage kidney disease; IQR, interquartile range.
[a]$P < 0.05$ by Chi Square or Wilcoxon testing.

Table 2. Analysis of no show to RGC appointment(s) or failure to submit specimens for genetic testing

Factor	Black (*n/N*)	White (*n/N*)	*P*-Value (χ^2)
No show to scheduled appointment(s) [Φa]	11.1% (6/54)	2.5% (6/238)	0.006
Failure to submit samples	14.5% (7/48)	4.3% (10/232)	0.01
Combined analysis [Φa]	24.1% (13/54)	6.7% (16/238)	0.0005

RGC, Renal Genetics Clinic.
Monte Carlo simulation was utilized to account for small sample size.
[Φ]indicates 12 additional patients (6 Black, 6 White) who scheduled but did not show up for RGC appointment(s) were included in this analysis only.

When analyzing data on "no show" to scheduled adult General Nephrology Clinic appointments on the main campus of Cleveland Clinic among a total of 38,589 patients, including 11,695 Black patients and 26,894 White patients, Black patients were also more likely to have "no show" compared with White patients (24.3% vs. 9.0%, $P = 2.2\text{x}10^{-16}$). Furthermore, Black patients were more likely not to submit samples despite the verbal affirmation of a decision to undergo genetic testing and RGC patient outreach (14.5% vs. 4.3%, $P = 0.01$). Patients who failed to submit samples were contacted by telephone calls, voice messages, and MyChart messages. White patients received outreach communications a median of 2.5 times following decision to undergo testing whereas Black patients received outreach communications a median of 2 times ($P = 0.28$).

The provider referral distribution to the RCG were similar among Black and White individuals, respectively: adult nephrology (63% vs. 54%, $P = 0.32$); pediatric nephrology (13% vs. 14%, $P = 0.93$); primary care providers (13% vs. 9%, $P = 0.59$); obstetrician-gynecologist (2% vs. 3%, $P = 1.00$); geneticists (2% vs. 3%, $P = 1.00$); rheumatologists (2% vs. 5%, $P = 0.7$); and self-referral (2% vs. 3%, $P = 1.00$).

Genetic Testing Modality

In the vast majority of Black and White individuals referred, genetic testing was ordered (98% vs. 94%, respectively) with a small percentage not ordered because of low index of suspicion (2% vs. 6%). As shown in Figure 1, multigene panel was the most frequently used genetic testing modality in Black patients (70%), followed by single gene test (30%), ES (23%), and chromosomal microarray (15%). No statistically significant differences in genetic testing modality were identified across racial groups.

Diagnostic Yield

Of the 47 Black patients who were recommended to have genetic testing, 35 had results available for analysis at the time of this manuscript submission. As shown in Figure 2, of those with results available, 13 (37%) had positive results; 4 (11%) had 2 *APOL1* risk alleles; 4 (11%) had heterozygous pathogenic variant for a phenotype-related autosomal recessive disorders (carrier); 7 (20%) had variants of uncertain significance; and 6 (17%) had negative results. No statistical difference in diagnostic yield between Black and White patients was observed (37% vs. 42%, $P = 0.69$). As shown in Table 3, the vast majority of genes only have 1 patient affected in this group, with *PKD1* and *GLA* genes having 2 patients each. The phenotypic characteristics and genetic testing findings among Black patients with positive testing results were listed in the Supplementary Table S1.

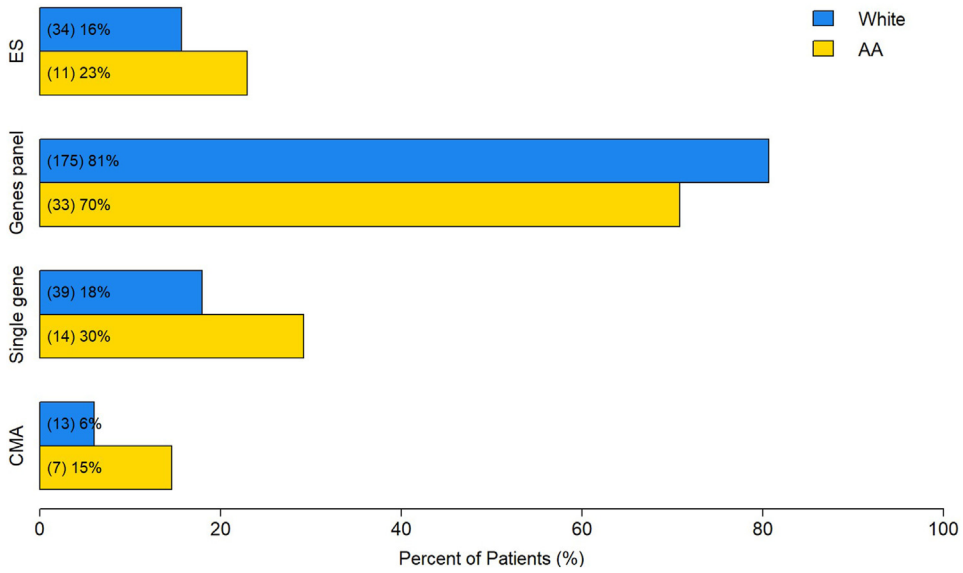

Figure 1. Suggested genetic testing modality in Black versus White patients in the Renal Genetics Clinic. Analysis was performed among Black and White patients who were suggested to have genetic testing(s). CMA, chromosomal microarray; ES, exome sequencing.

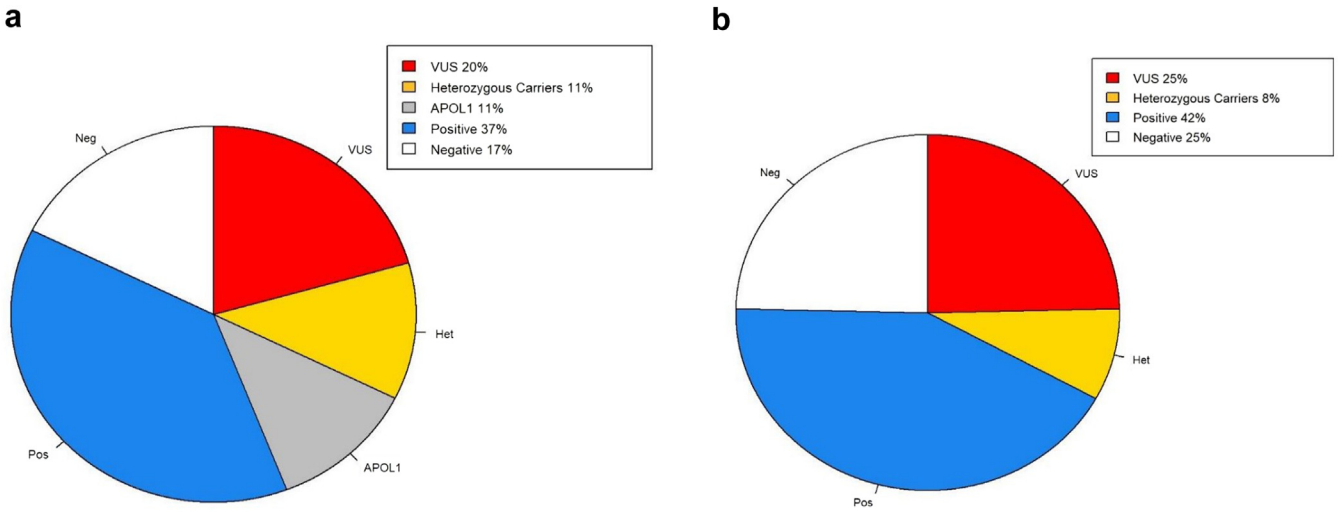

Figure 2. Diagnostic yield of genetic evaluation in Black patients (a) and White patients (b). Analysis was performed among Black and White patients who had genetic testing results available. APOL1, positive for 2 APOL1 risk alleles; Heterozygous (Het), heterozygous carriers; Negative (Neg), negative result; Positive (Pos), pathogenic, or likely pathogenic test results; VUS, variants of unknown significance.

As shown in Figure 3, the majority of positive results in Black patients were identified by multigene panels (7 out of 13, 54%), followed by single gene test (4 of 13, 31%) and ES (2 of 13, 15%). No statistically significant difference in diagnostic yield was noted between Black and White patients for any specific genetic testing modality.

Impact on Diagnosis and Management

As shown in Table 3 and Supplementary Table S1, among 13 Black patients with positive results, 9 unique monogenic disorders and 1 chromosomal microdeletion syndrome were diagnosed, compared with 39 monogenic disorders and 2 microdeletion syndromes out of 86 White patients with positive results.

The impact of genetic testing result on diagnosis and management are listed in detail in Supplementary Table S1. As shown in Table 4, with the genetic evaluation, 69% (9 of 13) Black individuals with positive results received a new diagnosis or a change in diagnosis. *A priori* diagnoses were confirmed or not changed in 31% (4 of 13) of patients. The new or changed diagnosis among patients led to a change in major management defined as initiation or discontinuation of medications before this manuscript submission in 44% (4 of 9) of patients. No difference in the implications of diagnosis and management was noted in Black and White patients (69% vs. 64%, $P = 0.77$; 44% vs. 40%, $P = 1.00$).

Table 3. The list of genes and/or chromosomal anomalies identified in Black patients who received positive testing results

Gene(s)	OMIM#	Number of Patients
PKD1	601313	2
CLCNKB	602023	1
CFHR1[a]	134371	1
CFHR1-CFHR3[a]	605336, 134371	1
GLA[a]	300644	2
7q31.1 deletion	602081	1
KMT2D	602113	1
COL4A5[a]	303630	1
INF2	610982	1
LCAT[a]	606967	1
ALPL	171760	1
HNF1B[a]	189907	1

[a]Indicates gene was identified in patients with ESKD on dialysis or status post kidney transplant.

DISCUSSION

In this study, we analyzed the clinical characteristics and genetics evaluation among Black patients in the RGC and identified several important findings. First, Black patients were more likely to be with ESKD at the time of referral compared with White patients. Second, Black patients were more likely to be covered by Medicaid and less likely to be covered by private insurance when compared with White patients. Third, Black patient were more likely to "no show" to scheduled RGC appointment(s) or not submit specimens of genetic testing compared with White patients. Finally, Black patients equally benefit from renal genetic evaluations with improved diagnosis and management compared with White patients.

Early diagnosis is critical for the management and outcome of patients with genetic kidney diseases. However, late diagnosis is not uncommon, which many attribute to the rarity and complexity of genetic disorders. Racial bias and delayed diagnosis have been noted in children with genetic conditions.[22] With more therapeutics for genetic conditions available, the need for timely and equitable genetic diagnosis has become more

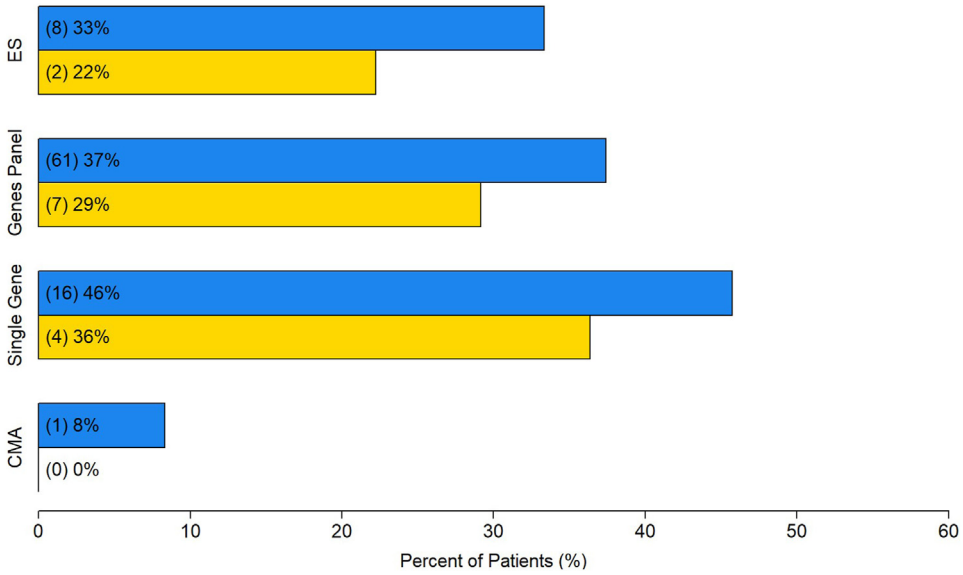

Figure 3. Diagnostic yield by testing modality in Black versus White patients. Number and percent of positive results per patient who received testing via each individual modality reported. Analysis was performed among Black and White patients who had genetic testing results available. CMA, chromosomal microarray; ES, exome sequencing.

important. In this study, participants were referred to the RGC from a variety of providers for a broad range of clinical presentations. We observed a higher proportion of Black patients were with ESKD at the time of referral compared with White patients, indicating late access of Black patients to genetics evaluation.

Further studies are required to investigate specific barriers for late access from both referring physicians and patients' perspectives. Factor(s) driving late access of Black patients to genetic evaluations are likely multifaceted and related to overall disparities in CKD outcomes in Black patients.[11,23] Socioeconomic inequity is one of these important factors. In this study, we observed that type of insurance coverage differed significantly between Black to White patients with kidney disease. Black patients were significantly more likely to be covered by Medicaid, and less likely to be covered by private insurance. Usually, individuals covered by public insurance (i.e., Medicaid) disproportionately encounter barriers to coverage approval for genetic testing when compared with individuals covered by private insurance.[24] It is encouraging that some Clinical Laboratory Improvement Amendments-certified laboratories, such as GeneDx and Natera, are offering genetic testing with zero out of pocket cost for patients with Medicaid regardless of insurance coverage.

In this study, all patients with Medicaid were offered free sponsored testing. However, we still observed that Black patients were more likely not to submit the specimens despite the decision to undergo genetic testing and RGC patient outreach. Ideally, patient samples would be collected at the time of initial evaluation. However, many patients must wait for insurance preauthorization to go through or apply for sponsored testing before samples can be collected. For patients who are able to or willing to pay out of pocket for genetic testing, samples can be collected on the same day. Therefore, the insurance preauthorization process adds a barrier for patients of lower socioeconomic status to submit samples. We hypothesize that this barrier contributes to the higher rate of failure to submit samples observed among Black patients. This indicates that an evaluation of educational strategies for conveying the importance and improving accessibility of genetic evaluation in the diagnosis and management of kidney diseases is necessary.

"No shows" (missed appointments) have been linked to adverse outcomes known to affect racial or ethnic minorities.[25,26] In this study, we observed a higher "no show" rate among Black patients in the RGC, with a similar trend noted in the General Nephrology Clinic. This suggests that the genetics clinic itself may not be the primary reason for the higher "no show" rates

Table 4. The diagnostic and management implications among Black vs. White patients who received positive results. One White patient had both confirmed *a priori* diagnosis and a new diagnosis

Impact	Black patients ($N = 13$)	White patients ($N = 86$)	*P*-Value (Chi Squared)
New diagnosis	8 (61%)	52 (60%)	1.00
Change of diagnosis	1 (8%)	3 (3%)	1.00[a]
Confirmation or no change of a prior clinical diagnosis	4 (31%)	32 (37%)	0.78[a]
Change in management[b]	4 (44%)	22 (40%)	1.00[a]

[a]Indicates Monte Carlo simulation was utilized to account for small sample size.
[b]Indicates analysis was performed only among those with new diagnosis or change of diagnosis.

among Black patients and that other factors involving a broad range of social determinants of health may contribute to this disparity. Interestingly, we observed a lower "no show" rate among both Black and White patients in the RGC compared to the General Nephrology Clinic. It may reflect the strong motivation of patients to pursue a genetic diagnosis given the complexity of their disorders and concerns for the risks of their family members. Studies including patients' surveys will be needed to understand the reasons for "no shows" and address the barriers for access to RGC.

Notably, we observed differences in males and females among Black patients compared with White patients with kidney disease. Whereas a higher proportion of White patients seen at the RGC were women, the opposite is observed among Black patients. Whereas males comprised only 33% of White patients with kidney disease seen at the RGC, 56% of Black patients with kidney disease were male ($P = 0.003$). Broadly, it has been observed that prevalence of CKD is higher in women compared to men. However, more men than women initiate kidney replacement therapy. One study found that CKD awareness is lower among US women compared with men.[27] In another study, it was determined that themes, including commitment to caregiving, deprioritizing own health, centrality of family in decision-making, self-reliance, and avoidance of placing burden on family may contribute to lower utilization of renal replacement therapy among women.[28] It has not been investigated whether this association varies by race or ethnicity.[27-29] However, we may hypothesize that if societal expectations for women are influencing utilization of health care, then this effect will be compounded by racism. Further investigation is required.

ES has been a powerful diagnostic tool for genetic disorders. However, due to its high cost, it has not been routinely used as first-tier clinical test except in cases with remarkable complexity and high suspicion for rare disorders. In this study, ES was recommended more commonly in Black patients compared to White patients though the difference did not reach statistical significance. It may indicate that Black patients were referred for genetic evaluation with more complicated medical histories and/or higher indices of suspicion for genetic disorders, even though they may experience late access to renal genetics evaluation.

G1 and G2 variants in the *APOL1* gene increase the risk for multiple subtypes of kidney disease, including focal segmental glomerulosclerosis and hypertensive kidney failure. *APOL1* risk alleles are increasingly important in disease management, particularly for patients undergoing living donor evaluation. These variants have an estimated prevalence of 13% in individuals of recent African ancestry,[5(p1),14(p1)] which is higher than that identified in this study (11.1%, 4 out of 35). This is likely because some patients were not tested for *APOL1* if they presented with nonglomerular disorders such as polycystic kidney diseases. Importantly, small molecule therapeutics targeting *APOL1* nephropathies are in development.[5(p1),30(p1),31] Therefore, genetic testing is particularly important for Black patients with kidney diseases and it may provide future opportunities for precision therapy.

Variants of uncertain significance is a common finding in genetic testing, as noted in our previous study. At the RGC, we adhere to the guidelines provided by the American College of Medical Genetics, which states that "A variant of uncertain significance should not be used for clinical decision-making. Efforts to reclassify the variant as pathogenic or benign should be pursued. While the variant is being reclassified, it may be prudent to monitor the patient for the associated disorder."[19] If a patient is identified with a variants of uncertain significance that overlaps with their phenotype, we conduct familial variant(s) tests for segregation analysis and functional studies (if available) to assist in the reclassification of the variants of uncertain significance .

This study has several limitations. Although this study has one of the largest groups of Black individuals in evaluation of an RGC, we recognize that the group size of Black patients is relatively small. We expect that an increased sample size of Black patients with kidney disease will improve the strength of associations noted in this study. Second, this is a retrospective cohort study from a single academic center. Further, small sample size limited the evaluation of other racial groups. Future work should incorporate multiple centers with renal genetics clinics to both increase sample size and assess for geographic differences in observations. Third, we did not have data surveying patients experience. We suggest further studies to evaluate Black patients' attitudes toward genetic testing and patient education practices are required.

In summary, this study suggests that Black patients with kidney disease equally benefit from genetics evaluation compared with White patients. Black patients benefit from improved diagnosis and management following genetic evaluation. However, substantial barriers in access to genetic testing exists, evidenced by the following: late access, higher "no show" rate to RGC appointments, and higher likelihood of not submitting specimens for genetic testing. As genetic testing and personalized genetic health care becomes increasingly central to clinical management in patients with kidney diseases, further studies are needed to investigate the barriers from

both patients' and physicians' perspectives to ensure equitable access for all persons.

DISCLOSURE

CG reports receiving research funding from NIDDK and serves as American Society of Nephrology Councilor. XW is a scientific advisory board member of Natera. M-BR reports receiving consulting fee from Alexion. JS reports receiving grants from NIDDK, Sanofi, and Vertex, as well as grants from NIAID, outside the submitted work. In addition, JS has a pending patent invention disclosure and serves as the Chair of the Steering Committee for KidneyX.

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SUPPLEMENTARY MATERIAL

Supplementary File (PDF)

Table S1. Phenotypic and genotypic characteristics of Black patients with positive test results and their implications on diagnosis and management.

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