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Views Of Primary Care Providers On Testing Patients For Genetic Risks For Common Chronic Diseases

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Abstract

We surveyed 488 primary care providers in community and academic practices in New York City in the period 2014–16 about their views on genetic testing for chronic diseases. The majority of the providers, most of whom were current or recent physicians in training, had had formal genetics education and had positive views of the utility of genetic testing. However, they felt unprepared to work with patients at high risk for genetic conditions and were not confident about interpreting test results. Many were concerned that genetic testing might lead to insurance discrimination and lacked trust in companies that offer genetic tests. These findings point to some of the attitudes and knowledge gaps among the providers that should be considered in the clinical implementation of genomic medicine for chronic conditions. Enhanced training, guidelines, clinical tools, and awareness of patient protections might support the effective adoption of genomic medicine by primary care providers.

Translation of genomic discoveries into routine clinical care will expand with growing knowledge of genetic variants; their impact on various diseases and treatments; and the availability of high-throughput, lower-cost genomic technologies.[1] Advances have led to genotype-directed clinical trials that have validated genomic medicine, clear

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recommendations by experts and professional associations, third-party reimbursement for increasing numbers of genetic tests,[2,3] standardization of formats for returning genetic results in electronic health records (EHRs), and use of gene-guided clinical decision support tools.

Genetic testing has been the purview of specially trained genetic medicine physicians and counselors. As testing becomes more commonplace and affordable, more clinicians will be able to incorporate genetic test results into their diagnostic and therapeutic arsenals. This is particularly true for primary care providers, who are responsible for comprehensive assessments of their patients' risks for common chronic diseases and choosing appropriate pharmacologic therapies. Genetic testing is increasingly relevant for these key aspects of primary care practice.[4,5] Widespread adoption of genetic medicine by these providers will depend, in part, on clinicians' buying in to testing and their ability to interpret and act on relevant genetic information.[6,7]

Studies to date have found that primary care providers report limited knowledge of and confidence in interpreting genetic test results and explaining them to patients.[8–12] Most studies have focused on the providers' general understanding of and preparedness for implementing genetic testing. Little is known about their specific preparedness to use genetic testing for common chronic disease risk, diagnosis, and therapy.[13–17] Communicating and acting on genetic risk for chronic disease is likely to be more complicated than pharmacogenomic testing to guide therapy, as it might require nuanced discussions with patients about disease risks they might not be aware of, lifestyle changes, and increased disease surveillance.

The implementation of chronic disease genetics is further complicated by increased disease-related morbidity and mortality among racial and ethnic minority groups.[18,19] There are few data on primary care providers' perceptions about genetic testing in diverse populations or their views about identifying and addressing variants more common among certain ancestral subgroups.[20–22] The promise and peril of genetic testing in diverse populations constitute an emerging area of exploration, on one hand making a clear distinction between race (a social construct) and ancestry (which has biological elements), and on the other hand raising genuine concerns that genetic testing could deepen racial stereotypes and stigmas.[23–26]

Our community-clinical-academic team developed a randomized clinical trial called Genetic Testing to Understand Renal Disease Disparities to test adults of African ancestry for high-risk *APOL1* renal disease variants, return test results to patients and their primary care providers, and assess the impact of this process on clinical care and patient outcomes.[27,28] The presence of two *APOL1* risk variants, nearly exclusively found in people of African ancestry, is associated with a tenfold increased risk for hypertension-related kidney failure.[29]

Immediately before enrolling their patients in the trial, we surveyed primary care providers to assess their attitudes and beliefs, generally about genetic testing and specifically for common chronic diseases. Our objective was to generate insights for the sustainable adoption and large-scale dissemination of genomic medicine, both broadly and for diverse clinical settings and ancestral populations.We hypothesized that primary care providers—

even if more recently trained—would feel unprepared to incorporate genetic testing for common chronic diseases in patient care and that they would have concerns about links between health disparities and genomics.

Study Data And Methods

We developed a forty-five-item, five-minute survey based on medical literature; the results of open-ended interviews with fifteen primary care providers; and discussions with the study's genomics board comprising patients, community advocates, clinicians, and researchers.[30] The survey asked providers about a number of their perceptions regarding genomic medicine, including associations between ancestry and genetic risk for common diseases, perceived knowledge and expertise about genetics, the clinical utility of genomic medicine, and their own genetics education.[28]

During regularly scheduled meetings in the period 2014–16, we asked primary care providers at four academic and six community-based general internal medicine practices (primarily staffed by internists) and five federally qualified health centers (primarily staffed by family physicians) in New York City to consider completing the survey as part of the process of educating them about the Genetic Testing to Understand Renal Disease Disparities study and obtaining their consent. We invited those with missing paper surveys to complete them online, and we provided no incentives. The providers included general internists, family physicians, nurse practitioners, physician assistants, and residents. Some of the practices chiefly served patients of African ancestry, while others chiefly served patients of European ancestry. Some primarily served low-income patients with public insurance, and others primarily served patients with private insurance. The Institutional Review Boards at all participating sites approved the project.

DATA ANALYSIS

The study team used descriptive analyses for primary care providers' demographic characteristics, means or standard deviations for continuous variables, and frequencies or proportions for categorical variables.We compared responses between subgroups of providers by clinical setting (academic or community) and by race/ethnicity, medical training status, and years in practice, using chi-square tests. We assessed differences in views using multivariate logistic regression, adjusting for testing experience and clinical setting. Our analyses were conducted using SAS, version 9.4. Significance was set at the 0.05 level. We combined black and Hispanic primary care providers in analyses of racial/ethnic subgroups because of small sample sizes for these respondents, along with similar responses.

LIMITATIONS

This study had several limitations. First, the survey was conducted in one large city with a primary care provider population that consisted predominantly of physicians in training or recent medical graduates, and therefore the results might not be broadly generalizable. Second, the survey was a pre-implementation study: Primary care providers' attitudes could change after their patients were tested.

However, these limitations should be balanced by the study's strengths, which included surveying nearly 500 diverse providers from a variety of settings, having a high response rate, and asking providers to respond to testing that their patients were about to receive rather than to a hypothetical scenario.

Study Results

Of the 547 primary care providers invited to participate, 488 (89 percent) completed the survey. Most had been in practice for less than five years, were female, were medical residents, and practiced in an academic setting (exhibit 1). Half self-identified as white. Nearly all considered patient care to be their primary job.

VIEWS ON GENETIC TESTING

Most of the providers had a positive view of genetic testing for common chronic diseases: 74 percent agreed that it is clinically useful, and 70 percent agreed that it will improve clinical outcomes within the next five years (exhibit 2). Just over half believed that their patients would be interested in genetic testing for chronic diseases. One-third believed that testing would motivate their patients to adopt healthy behaviors.

PREPARATION FOR GENETIC TESTING

Most primary care providers had had some formal genetics education (exhibit 2). Yet only one-third had ordered any genetic test, returned a genetic test result to any patient, or referred a patient for genetic counseling in the past 12 months (exhibit 3). Surprisingly, past testing experience was not associated with provider attitudes toward or confidence in interpreting genetic testing results. Despite their general genetic training and experience, only 40 percent of primary care providers felt knowledgeable about the genetic basis for common diseases. And few felt prepared for working with patients who have had genetic testing for common diseases (25 percent) and those at high risk for genetic conditions (28 percent). Only 14 percent felt confident about interpreting test results.

CONCERNS ABOUT GENETIC TESTING

One in two primary care providers were concerned that genetic testing would lead to insurance discrimination (exhibit 2). Three out of four mistrusted companies that offer genetic testing, four out of five thought that insurance wouldn't cover the cost of genetic testing in five years, and over half believed that telling patients their genetic risk for chronic disease could cause them excessive stress (exhibit 3).

RACE, ANCESTRY, DISPARITIES AND GENETICS

Eighty-one percent of primary care providers agreed that genetic variants provide clues about causes of racial and ethnic disparities and that race or ancestry can identify patients who would benefit from genetic screening for diseases (exhibit 3). Seventy-five percent considered patients' race or ancestry when making decisions about which medications to prescribe (exhibit 2).

RESOURCES THAT PRIMARY CARE PROVIDERS WANT

Over 80 percent of the providers wanted an easy way to order genetic tests, more information on patient management when tests were positive, and handouts to give their patients on genetics and genetic testing in general, as well as specific information on positive tests (data not shown). Nearly 70 percent wanted information on how to talk with patients about genetics and genetic testing.

DIFFERENCES IN PRIMARY CARE PROVIDER SUBGROUPS

We found no significant differences between primary care provider subgroups (those in academic versus community clinical settings, whites versus nonwhites, and by number of years in practice) in their self-reported preparedness to use genomics in practice and views on genetics, or in the resources they wanted. However, providers in the black or Hispanic group (odds ratio: 2.31) and Asian providers (OR: 2.04) were more likely than whites to agree that genetic testing would motivate patients to adopt healthy behaviors ($p = 0.001$, OR adjusted to test experience) (exhibit 3 and online appendix exhibit A).[31] There were similar differences between white and nonwhite providers about whether discussing genetic risk for kidney disease with patients would delay or prevent its onset (black or Hispanic OR: 3.49, Asian OR: 1.78; $p < 0.05$; OR adjusted for ordering test experience and clinical setting) (exhibit 3 and appendix exhibit B).[31] And nonwhite providers were more likely than whites to agree that providing genetic risk information would cause patients excessive worry or stress.

Academic clinicians (mainly internists) were less likely than community clinicians (mainly family physicians) to have ordered a genetic test in the past 12 months (24 percent versus 56 percent), returned results to a patient (18 percent versus 52 percent), or referred a patient for genetic counseling (23 percent versus 63 percent) (exhibit 3). Academic clinicians were significantly more likely than community clinicians to believe that genetics, in part, explains health disparities (85 percent versus 74 percent). Community clinicians were more likely than academics to agree that risk information would cause excessive worry (65 percent versus 55 percent).

Compared to primary care providers with five or more years in practice, those with less time in practice were more likely to think that genetic medicine would improve clinical outcomes (75 percent versus 62 percent; $p = 0.02$) and less likely to be concerned that sharing genetic risk information would cause patients excessive stress (54 percent versus 68 percent; $p = 0.002$) (data not shown). Similarly, compared to attending physicians, fellows, nurse practitioners, and physician assistants, residents were more likely to think that genetic medicine would improve outcomes (73 percent versus 63 percent; $p = 0.003$) and less likely to be concerned that risk information would cause patients excessive stress (52 percent versus 68 percent; $p = 0.003$).

Discussion

The expansion of genomic medicine into primary care will depend on the preparedness of clinicians to incorporate into practice the information gained, particularly as findings

become more relevant to patients from diverse backgrounds and those with or at risk for common chronic diseases. Our survey indicated that most primary care providers have had some formal genetics education, have positive views of genetic testing's utility, and think that their patients would be interested in having testing but feel unprepared to work with patients at high risk for genetic conditions or who have had genetic testing. Primary care providers' belief that genetic testing would motivate patients and prevent disease was countered by concerns about patients' negative reactions to their results and insurance discrimination (despite little evidence that genetic discrimination occurs), as well as mistrust of companies that offer testing.[32]

The differences in experience and perceptions that we identified among subgroups of primary care providers merit further exploration. Providers in community sites had more experience ordering testing and returning results, likely because nearly all were family physicians who ordered prenatal tests, whereas academic providers were nearly all internists who did not order these tests.

It is interesting that most primary care providers linked genetics and disparities, and that nonwhite providers were more optimistic than whites that their patients would change their behaviors and benefit from testing. In one earlier study, black physicians more commonly indicated that patient race is a central factor for choosing among treatment options and understanding disease risk;[20] this was not borne out in our larger quantitative study. Another study found few differences between blacks and whites in terms of attitudes, but it did find that physicians in general were reluctant to connect genetics, race, and disease and believed that genetics had a limited role in explaining racial differences in health.[33] Our more recent study showed less reluctance and more positive views of the role of genetics in medicine, even in a diverse primary care provider population with a very high response rate. More research should be conducted to corroborate our finding that nonwhite physicians were more optimistic than whites about the impact of genetic testing on patient behaviors and outcomes. Genetic Testing to Understand Renal Disease Disparities and other implementation trials will determine whether this optimism is merited.

Policy Implications

Even among primary care providers who stated that they had had formal genomics education and who had recently been trained (in an era characterized by an explosion in genomic discoveries and technology), perceived preparedness was low. Interest in, enthusiasm for, and experience with genetic testing were not associated with confidence in working with patients who had genetic testing for common chronic conditions. Therefore, improving primary care providers' skills will require more than additional training and experience. It will likely also require developing and deploying systems to facilitate testing and the returning of results, as well as to provide information for providers and patients at the point of care. Most of the providers we surveyed wanted to use the EHR to order genetic tests and obtain genetics-related information for themselves and their patients.[27] Emerging systems are testing clinical decision supports in EHRs to deliver actionable genetic information to primary care providers,[34] which could provide support for providers at all levels of genetics education and experience.

Professional societies are also developing guidelines to ensure the use of appropriate genetic testing, educate their members,[35] and improve providers' competency in genomic medicine.[36,37] These resources can and should be usable for primary care providers.While the factors involved in preparing physicians for the integration of genomics into practice are complex, the introduction of other medical innovations has raised and overcome similar challenges, and promoting "genomic exceptionalism" by limiting its use to specialists may be unwise.[38]

Despite having positive views about the utility of genetic testing, the clinicians in our study doubted that testing would motivate patients to adopt healthy behaviors. They might have been drawing on their experience and evidence from meta-analyses of risk-informed behavior change that found little evidence that disclosure of genetic risks catalyzes health-related behavior change.[39–41] Risk information in these studies was generally not coupled with support for behavior change, which could have limited its effectiveness. The optimal use of genetic information related to common chronic disease risk will likely need to draw on evidence-based methods of promoting or facilitating behavior change and providing tools in EHRs for primary care providers to help their patients.

It will also be important to address providers' concerns, such as about insurance discrimination resulting from genetic testing. While there are legal protections in place, notably the Genetic Information Nondiscrimination Act (GINA) of 2008, clinicians and patients are not sufficiently aware of these provisions or their benefits and limitations.[42] Greater understanding of patient protections should be part of primary care providers' education, especially as the legal landscape around genetics evolves.[43] Providers reported a lack of preparedness for genetic medicine, and mixed experiences with direct-to-consumer testing might have played a role in their perceptions. The impact of genetic testing on patients' insurance coverage, costs, and access to care must be carefully monitored.[44]

Our study also explored associations between race/ethnicity and genetics—an important but understudied field.[30] We found that despite concerns about linking race, ancestry, and genetics, primary care providers of all backgrounds believed that health disparities have a genetic basis.[45,46] It will be important to determine how to use genetic discoveries for the benefit of all patients and to address the significant underrepresentation of minority populations both in genetics research[47,48] and as partners in the development and implementation of translational research, policy, and practice.[49] Fortunately, research networks are beginning to close research gaps, reduce racial and ethnic disparities in the use of currently available tests for genetic disease risks, and use findings to improve outcomes.[50]

Conclusion

Primary care providers are key stakeholders in the adoption of genetic medicine.[51] A majority of the providers we surveyed believed in the potential clinical benefits of genetic testing for common chronic diseases but felt that they lacked the knowledge and skill to use genetics in the care of their patients. Efforts to expand genetic testing should help identify common, actionable variants that increase chronic disease risk and should enhance primary care provider training and the use of EHRs to help clinicians act on the increasing volumes of genetic information they will encounter. Further studies of the impact of such strategies

and initiatives to ensure that community practices and low-income and minority populations have access to new discoveries from which they can benefit will likely increase the successful adoption of genomic medicine into practice.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

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It will be important to address providers' concerns, such as about insurance discrimination resulting from genetic testing.

EXHIBIT 1

Demographic characteristics of primary care providers who completed the genetic testing knowledge and viewpoint survey

Characteristic	Number	Percent
Female	290	64
RACE/ETHNICITY		
White	252	52
Black	34	7
Hispanic	34	7
Asian or South Asian	139	28
Other	29	6
POSITION		
Physician (attending) or fellow	147	31
Nurse practitioner	21	4
Physician (resident)	313	65
YEARS IN PRACTICE[a]		
0–5	371	77
>5–10	35	7
>10–15	18	4
>15–20	21	4
>20	34	7
CLINICAL SETTING		
Academic	309	64
Community	175	36
SPEND MAJORITY OF TIME ON PATIENT CARE		
Yes	433	91

SOURCE Authors' analysis of survey data from 2014–16. **NOTES** $N = 488$. The mean age of the providers was 33.3 years, and the standard deviation was 9.5 years.

[a]Percentages do not add up to 100 because of rounding.

EXHIBIT 2

Primary care providers' views about and perceptions of genetic testing

View or perception	Percent
AGREED OR STRONGLY AGREED THAT:	
Genetic testing for common disease risk is clinically useful	74
Within five years, genetic medicine will improve clinical outcomes	70
My patients would be interested in genetic testing for common diseases	53
Genetic testing would motivate my patients to adopt healthy behaviors	34
Genetic variation provides clues to causes of racial/ethnic disparities	81
AGREED OR STRONGLY AGREED THAT THEY:	
Are knowledgeable about the genetic basis of common diseases	40
Are ready to take care of patients who had genetic testing for common diseases	25
Are prepared to work with patients at high risk for genetic conditions	28
Are confident interpreting genetic test results	14
Have had formal genetics education	78
Are concerned that genetic testing will lead to insurance discrimination	53
Trust companies that offer genetic testing	24
Consider race/ancestry when deciding which medications to prescribe	75

SOURCE Authors' analysis of survey data from 2014–16. **NOTE** There were 488 providers.

EXHIBIT 3

Variations in primary care providers' perceptions of genetic testing, by race/ethnicity, clinical setting, and position

	Percent of providers							
		Setting		Race/ethnicity			Position	
	All (N = 488)	Community (n = 175)	Academic (n = 309)	White (n = 252)	Black or Hispanic (n = 68)	Asian (n = 139)	Resident (n = 313)	Attending, fellow, or NP (n = 168)
AGREE OR STRONGLY AGREE THAT:								
Genetic testing will motivate my patients to adopt healthy behaviors	34	29	37	27**	45**	43**	32	37
Discussing genetic risk will delay onset of disease	46	46	47	38**	68**	53**	45	48
Testing will improve patient outcomes	70	62**	75**	69	75	73	75**	62**
Testing may lead to insurance discrimination	53	61**	49**	50	55	60	52	57
In five years insurance will not cover cost of genetic testing	80	82	80	83	72	79	79	83
Telling a patient they have genetic risk for chronic disease may cause excessive stress	59	65**	55**	52**	65**	64**	54**	68**
Don't trust companies offering testing	76	83**	72**	76	72	76	74	89
Race/ancestry can identify patients who benefit from disease screening	81	74**	85**	85	76	80	83	79
Genetic variations provide clues to causes of racial/ethnic disparities	81	74**	85**	84	79	81	82	81
IN THE PAST 12 MONTHS:								
Ordered a genetic test for any patient	36	56**	24**	35	39	35	28**	50**
Returned genetic test results to any patient	30	52**	18**	30	37	27	22**	46**
Referred a patient for genetic counseling	38	63**	23**	38	41	37	23**	66**

SOURCE Authors'analysis of survey data from 2014–16. **NOTES** Categorical sample sizes do not equal the total sample size because of missing values. Significance refers to differences within categories. NP is nurse practitioner.

** $p < 0.05$