Evaluating Primary Care Providers' Readiness for Delivering Genetic and Genomic Services to Underserved Populations

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Keywords

Genetics · Genomic services · Primary care · Underserved

Abstract

Introduction: Increased genomics knowledge and access are advancing precision medicine and care delivery. With the translation of precision medicine across health care, genetics and genomics will play a greater role in primary care services. Health disparities and inadequate representation of racial and ethnically diverse groups threaten equitable access for those historically underserved. Health provider awareness, knowledge, and perceived importance are important determinants of the utilization of genomic applications. **Methods:** We evaluated the readiness of primary care providers at a Federally Qualified Health Center, the Community Health Center, Inc. (CHCI) for delivering genetic and genomic testing to underserved populations. Online survey questions focused on providers' education and training in basic and clinical genetics, familiarity with current genetic tests, and needs for incorporating genetics and genomics into their current practice. **Results:** Fifty of 77 (65%) primary care providers responded to the survey. Less than half received any training in basic or clinical genetics (40%), were

familiar with specific genetic tests (36%), or felt confident with collecting family health history (44%), and 70% believed patients would benefit from genetic testing. **Conclusion:** Despite knowledge gaps, respondents recognized the value and need to bring these services to their patients, though would like more education on applying genetics and genomics into their practice, and more training about discussing risk factors associated with race or ethnicity. We provide further evidence of the need for educational resources and standardized guidelines for providers caring for underserved populations to optimize appropriate use and referral of genetic and genomic services and to reduce disparities in care.

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Published by S. Karger AG, Basel

Introduction

Primary care has become the foundation of a strong health-care system, and has led to better health outcomes for many low socioeconomic communities [1]. However, access to primary care remains an ongoing challenge for many Americans, and disparities exist in primary care

clinics that predominantly serve minority communities, with less access to equipment, space, and specialists [2]. Many patients from low income, uninsured or Medicaid-insured, and/or racial and ethnic groups receive primary care services through community health centers, such as Federally Qualified Health Centers (FQHCs) [3]. FQHCs often serve multiple generations of families, throughout all stages of life, and for increasingly complex conditions [4, 5] and are leaders in providing comprehensive, culturally competent, and high quality primary health care. Many of these patients may face additional social health determinants that limit their abilities to gain access to specialty care, make informed decisions about testing, and understand and interpret genetic results independent of their providers due to low health literacy.

With the arrival of new precision medicine applications across health care, undergirded by the growth in genetics and genomics, primary care, and specialty care can benefit alike. Genomic information can help inform preventive strategies, management, and therapeutic decision-making [6]. Genomics is increasing knowledge on disease etiology, predisposition, prognoses, and treatment response, with demonstrated value and clinical importance of new diagnostics and interventions [7], throughout all stages of life [8, 9]. However, despite the increasing number of genomic tests and interventions, several factors limit integration of genomics into general practice settings, particularly provider preparedness [10–19]. Studies have also demonstrated lower referral rates and utilization of genetic applications by minority populations [20–24].

In addition, disparities in low participation of diverse groups in genetics and genomics can limit the clinical evidence basis [25]. The majority of genomic studies, which inform disease prediction and treatment, are conducted with those of European ancestry [26, 27], thereby limiting the predictive value of disease risk prediction and treatment efficacy for those of non-European ancestry. Increased diversity of these reference datasets will give providers stronger confidence in the care guidelines for predictive and preventive care informed by genomic data [28, 29]. As one of 6 FQHC recruitment sites for the National Institutes of Health's *All of Us* Research Program (AoURP), a component of the Precision Medicine Initiative® [30], our center is contributing increased diversity for future genomic studies and developing a more representative biomedical resource.

While much work has been conducted on assessing primary care providers' preparedness to incorporate genomics data in their practice [11–19], few have examined

providers and practices that serve primarily minority and vulnerable populations [31]. Many FQHC patients may be unable to comprehend and make informed decisions or access specialty care and thus require additional support from providers to benefit from genetics and precision medicine [32]. To assess primary care providers' serving racial and ethnic minorities, we surveyed providers' knowledge, perceived readiness and attitudes toward delivering genetic and genomic services. We report the findings of our assessment and discuss providers' needs in order to introduce genetics and genomics services for our patients, and suggest guidelines for other FQHCs.

Materials and Methods

Survey Development

We developed a 20-question survey to assess primary care providers' baseline knowledge and attitudes on incorporating genomics in their clinical practice. Survey development was guided by published work in this area [12, 33]. The questions covered 3 categories: (1) provider education and self-evaluated expertise; (2) knowledge of genetics, types of testing, and family health history; and (3) general attitude of providing this information for underserved patients. We asked about providers' awareness and level of comfort in discussing 6 specific types of genetic tests (carrier screening, diagnostic testing, newborn screening, pharmacogenetics, prenatal testing, and pre-symptomatic/predictive testing). Five answer choices were provided: (1) have not heard of this test; (2) have heard about this test, but am not prepared to discuss it with patients; (3) have heard but not offer any specifics; (4) have heard and am able to discuss with additional training, but not ready to implement in practice; and (5) have heard and am able to fully discuss and implement in practice. For the question about direct-to-consumer (DTC) testing, we assessed familiarity and attitudes through 5 response options: (1) no; (2) yes, but do not know of any specific tests; (3) yes, familiar with some specific tests; (4) yes, and would recommend to my patients; and (5) yes, and would not recommend to my patients. The Community Health Center, Inc. (CHCI) Institutional Review Board approved this study (#1155).

Survey Population

All primary care providers practicing at one of the 14 medical clinics at the CHCI based in Middleton, CT (USA), were eligible for this survey. The CHCI is an FQHC that delivers team-based care, with full integration of medical, dental, and behavioral health staff to deliver comprehensive, culturally sensitive and accessible health care within primary care center hubs across the state. In addition to traditionally delivered primary care services, the CHCI offers a variety of programs targeted directly at special populations. These include the Wherever You Are (WYA): Health care for the Homeless Program, mobile dental, school-based health centers, and Migrant Farm Worker Clinics to target patients in settings that are most convenient for them. Approximately, 76% of CHCI's patient population are minorities, over 89% are at or below 200% of the Federal Poverty level, 20% are uninsured, and over 66% are on Medicaid. Eligible primary care providers for this study

Table 1. Primary care provider knowledge base by specialty and average years of practicing

Provider	Specialty	Responses	Years practicing	Genetics training			Knowledge basic genetics				Knowledge clinical genetics					
				average	yes	no	do not recall	excellent	very good	good	fair	poor	excellent	very good	good	fair
NP (N = 25)	Behavioral health	2	0.75	2	0	0	0	1	0	1	0	0	0	0	1	1
	Family medicine	17	3.5	7	9	1	1	4	7	3	2	0	2	6	8	1
	Family medicine/ pediatrics	2	6.0	0	1	1	0	0	1	1	0	0	0	0	2	0
	Internal medicine	1	16.0	0	1	0	0	0	1	0	0	0	0	0	1	0
	Pediatrics	3	13.5	2	1	0	0	0	1	2	0	0	0	0	3	0
Physician (N = 22)	Family medicine	11	12.9	3	6	2	0	1	2	7	1	0	1	1	7	2
	Internal medicine	2	24.0	1	0	1	0	1	1	0	0	0	1	1	0	0
	Internal medicine/ Pediatrics	1	5.5	1	0	0	0	0	1	0	0	0	0	1	0	0
	Pediatrics	8	15.1	4	4	0	0	3	2	3	0	0	2	3	3	0
PA (N = 2)	Family medicine	1	30.5	0	0	1	0	0	0	0	1	0	0	0	1	0
	Pediatrics	1	9.0	0	1	0	0	0	0	1	0	0	0	0	1	0
Declined to answer (N = 1)	Unknown	1	30.0	0	1	0	0	0	0	1	0	0	0	0	1	0
NP, nurse practitioner; PA, physician assistant.																

included physicians, physician assistants (PAs), and nurse practitioners (NPs), regardless of their tenure at CHCI, for a total of 77 primary care providers.

Data Collection and Analysis

The survey was a self-administered, Web-based survey, through SurveyMonkey, with an average time to completion of 5 min. The survey was conducted between January 2020 and March 2020. A brief description of the project and an invitation to participate was sent via email from the chief medical officer with a link to the survey. The survey was open for 2 months and email reminders were sent every 2 weeks. The survey was anonymous, and no identifiable information was collected. Provider race and job title were not collected to protect participants' identity.

Data Analysis

We generated descriptive statistics for each survey question. Relationships between categorical variables were assessed with χ^2 testing. Calculated *p* values were considered significant at a type I error level of 0.05. All data were stored in an Excel file and analyzed using Excel.

Results

Respondent Characteristics

Of the 77 CHCI primary care providers (29 physicians, 46 NPs, and 2 PAs) invited to participate, 50 completed the survey, resulting in a 65% completion rate. Half (50%) were NP, 44% are physicians, and 4% PA (Table 1). Of the 50 respondents, 58% practiced in family medicine, 30% practice in pediatrics, 6% in internal medicine, and 2% in behavioral health. While the average number of years in practice for NPs was 5.2 years (median: 3.5 years; range: <1–25 years), a substantial proportion (20%) were new and have been practicing less than a year. In contrast, physicians and PAs have been practicing an average of 14.3 (range: 1–42) and 19.8 (range: 9 and 30.5) years, respectively.

Genetic Education and Knowledge Base

Providers were asked if they received any genetics training (undefined) and to rate their knowledge of basic and clinical genetics. A total of 40% received some genetics training (Table 1). Overall, 44% of NPs received genetics training, followed by 41% of physicians (41%). None of the PAs received any basic or clinical genetics training. We did not observe an association between number of years in practice and genetics training. Those NPs who had received some genetics training have been practicing from <1 year to 25 years, and represent the specialties of behavioral health, family medicine, and pediatrics. Of the physicians who received genetics training, more than half

have been practicing for 20 or more years. Of those practicing pediatrics, 47% (7/15) indicated some genetics training and 34% of family medicine (10/29) had some genetics training.

Overall, 22% of respondents (11/50) rated their knowledge of basic genetics as excellent or very good and 12% (6/50) as excellent or very good for clinical genetics. Of the 20 respondents who reported some genetics training, 55% rated their basic genetics knowledge as excellent or very good and 30% rated their knowledge of clinical genetics as excellent or very good.

Of the pediatric providers who had some genetics training (7/15), 43% (3/7) rated their knowledge of basic genetics as excellent or very good, and 28% (2/7) listed their knowledge of clinical genetics as very good (no providers rated their knowledge as excellent). Sixty percent of family medicine providers with some genetics training (6/10) rated their knowledge of basic genetics as excellent or very good, and 30% (3/10) rated their knowledge of clinical genetics as excellent or very good.

Familiarity with DTC Testing

Primary care providers were asked about their level of familiarity with DTC or patient-initiated personal genetic testing, such as 23andMe (<https://www.23andme.com/?mkbanner=true>) and Invitae (<https://www.invitae.com/en/patient-testing/>). The majority of respondents (92%) were familiar with DTC or patient-initiated genetic testing, though only 36% were familiar with specific tests. No trends were observed between familiarity with DTC and their role, genetics training or perceived level of knowledge for basic or clinical genetics.

To assess primary care providers' knowledge of clinical genetic tests, we asked them to rate their knowledge about 6 specific types of tests: carrier screening, diagnostic testing, newborn screening, pharmacogenetics, prenatal testing, and pre-symptomatic and predictive testing. Overall, the primary care providers indicated that they have heard of all of the tests (data not shown), and the majority of providers felt that they could discuss the tests but not offer any specifics. Providers were least likely to have heard of pharmacogenetics and newborn screening. Of all of the tests, providers were least able to discuss or ready to implement pharmacogenetics tests.

By specialty, 53% of those in pediatrics indicated they had the ability to discuss and implement newborn screening in their practice (data not shown). While all respondents had heard of diagnostic testing, only physician respondents were prepared to fully discuss and implement in their practice.

Table 2. Reported confidence with delivery of genetics and genomics services in current care practice

Provider	Specialty	Genetics in current practice			Confidence with collecting family health history				Identify red flags from FHH		
		no	yes, immediately	yes, with additional training	not at all confident	slightly confident	somewhat confident	moderately confident	very confident	no	yes, immediately
NP (N = 25)	Behavioral health	2	0	0	0	2	0	0	0	0	2
	Family medicine	13	1	3	1	2	8	4	2	0	10
	Family medicine/pediatrics	2	0	0	0	1	1	0	0	0	2
	Internal medicine	1	0	0	0	1	0	0	0	0	1
Physician (N = 22)	Pediatrics	3	0	0	0	2	1	0	0	0	2
	Family medicine	7	0	2	0	4	2	4	1	0	8
	Internal medicine	0	1	1	0	0	0	2	0	0	1
	Internal medicine/pediatrics	1	0	0	0	0	1	0	0	0	0
PA (N = 2)	Pediatrics	5	1	2	0	0	6	2	0	0	4
	Family medicine	0	0	1	0	0	1	0	0	0	1
Declined to answer (N = 1)	Pediatrics	1	0	0	0	0	1	0	0	0	1
	Unknown	1	0	0	0	0	1	0	0	0	1
NP, nurse practitioner; PA, physician assistant.											

Table 3. Providers' perceived benefits of genetic services to patients

Provider	Specialty	Benefit to your patients		Resources available		Patients asked		Patients interested	
		no	yes	no	yes	no	yes	no	yes
NP (N = 25)	Behavioral health	1	1	2	0	2	0	0	2
	Family medicine	5	11	7	10	10	5	3	14
	Family medicine/pediatrics	2	0	2	0	2	0	1	1
	Internal medicine	0	1	0	1	0	1	0	1
	Pediatrics	0	3	1	2	2	1	0	3
Physician (N = 22)	Family medicine	4	6	4	7	5	5	1	9
	Internal medicine	0	2	2	0	2	0	0	2
	Internal medicine/pediatrics	0	1	1	0	0	1	0	1
	Pediatrics	2	6	2	6	3	4	1	6
PA (N = 2)	Family medicine	0	1	0	1	1	0	0	1
	Pediatrics	0	1	1	0	1	0	0	1
Declined to answer (N = 1)	Unknown	0	1	1	0	0	1	0	1

NP, nurse practitioner; PA, physician assistant.

Bringing Genetics to Primary Care Practice

The final set of questions aimed at determining providers' attitudes and ability to incorporate genetics into their clinical care practice, specifically with respect to family health history and risk factors (Table 2). Seventy-five percent of providers (36/48) were not currently incorporating genetics or genomic medicine into their patient care practice and do not know how to incorporate genetics in their current care. Nineteen percent (9/48) felt that they could incorporate genetics in their current care with additional training.

With respect to family health history, 44% of providers indicated that they are "somewhat confident" with collecting family health history, 24% were moderately confident, and <1% were very confident. When asked about their ability to identify patterns or red flags in family health history for increased disease risk, 67% of the respondents responded "Yes, with some additional training," and the remaining 33% responded that they could now.

Providers were asked to rate their comfort level with discussing the following risk factors for common diseases with their patients: environment, genetics, health status, lifestyle, and race/ethnicity. Sixty-six percent of providers were comfortable currently with fully discussing risk factors associated with lifestyle, followed by health status (56%), environment risk factors (40%), race/ethnicity (34%), and genetics (16%). With more training, 40% indicated they would be comfortable discussing genetics, though 46% would like more training with discussing risk factors associated with race or ethnicity.

Seventy percent of respondents believed that patients would benefit from genetic testing (Table 3). A little over half (54%) responded that they did not have the resources needed to provide genetic testing. To facilitate delivery of genetic testing, the top resources identified by providers were more education on applying genetics to their practice, followed by insurance coverage and support staff. Regarding patient interest, 36% of providers indicated that they have had patients' inquire about genetic tests in the past year. Most providers (84%) thought their patients would be interested in genetic testing offered as part of their medical care.

Discussion

This work demonstrates that providers serving FQHC patients recognize the increased value of genetics and a role for genetic services in primary care. Primary care

provider attitudes and training are keys to the appropriate delivery of genomic medicine and integration of test results into care, and thus, continued education and training with genetics and genomics are critical [34]. In general, provider attitudes have been enthusiastic and recognize the importance of integrating genetic and genomic services into care, but several barriers to test utilization have been reported [14, 35]. A systematic literature review categorized these barriers around 4 themes: provider knowledge/skill, health-care system, ethical, legal, and social implications, and scientific evidence supporting use of genetic services [16]. Gaps with knowledge and skills were the most cited challenge. While the characteristics of the patient populations in prior surveys of providers' attitudes and knowledge about genetics are not typically reported, to our knowledge, there have been no publications on primary care providers caring for predominantly underserved populations. To reduce further widening of disparities regarding access and utilization of these technologies, it is critical to insure providers of underserved populations have the knowledge and tools to offer these genetics and genomic applications to their patients.

About 40% of providers surveyed reported having some genetics training; however, only 22% of respondents overall rated their basic knowledge of genetics as excellent and very good. An even smaller proportion (12%) rated their knowledge of clinical genetics as excellent or very good. There are likely differences in the content and emphasis on genetics/genomics between medical and nursing curricula, specialties, and training programs. Our data did find that more pediatric providers had genetics training (47%) than nonpediatric providers (38%). Prior studies are equivocal regarding the level of knowledge and utilization of genetic services [36–41], though pediatric providers may have more opportunities and exposure to genetics at professional meetings, clinical rotations, medical literature, and colleagues board-certified in pediatrics/medical genetics [42]. Furthermore, the nursing professional community has been very proactive in developing core competencies and educational programs in genetics dating back to the early 2000s [43–47], and the higher numbers with some genetic training is reflected in our data.

Respondents' low rating of their perceived knowledge of basic and clinical genetics contrasted with their reported familiarity with several types of genetic tests and comfort with discussing different types of testing with their patients. However, their almost unanimous response regarding their perceived inability to integrate genetics into

current practice indicates they are not fully prepared and indeed, they noted that they did not feel they had the adequate resources to offer and integrate test results into practice. In addition to low knowledge, other factors may also contribute to their perceived inability to integrate genetics into practice, such as lack of access/knowledge of genetics specialists and time constraints.

The FQHC providers did not express reservations about patient interest in genetic and genomics services and believed that genetic information could benefit their patients. However, it has been well documented that differences in the delivery and utilization of genetics and genomics technologies do exist, particularly for minority and ethnic patients [21, 48–50]. Patient concerns include fear of discrimination (even with federal protections) [51, 52] and medical mistrust [53]. Health providers that serve a large proportion of minority patients may have different concerns, clinical obligations, and resources available to them to offer genetic and genomic services than those working in settings with a smaller proportion of minority patients [2, 54]. The current practice of referral to outside specialists does not take into account other factors disproportionately affecting underserved patients, such as insurance access, time away from work, or transportation to the specialist clinic, and does not allow patients to play an informed and empowered role in their overall health. The patient population served at the CHCI FQHC is highly diverse with respect to every domain—age, race, ethnicity, linguistic preference, sexual orientation, and gender identity. Overall, these patients have higher rates of asthma, diabetes, depression, anxiety, substance abuse, and obesity. With the rapid expansion of genetic testing and increased value with treating chronic conditions, CHCI is looking to establish these services as the standard of care. Implementation of such standard would ideally align and integrate with existing clinical workflows and care delivery procedures. This would need to include embedding a screening process for patient risk that utilizes validated tools that are easy to score and interpret. Consideration must be given to integrating these tools into the electronic medical record. The roles of each staff member in the procedure of screening for genetic risk and follow up must be clearly identified, as well as identification of specialists for referral.

Interpretation of genetic susceptibility is further complicated by social health determinants and other biological factors, leading to a range of clinical symptoms/disease severity as reported in some minority populations, and in the FQHC setting [55]. Thus, training in basic and clinical genetics will not be sufficient to accurately incorporate ge-

netics data into risk prediction and treatment – current research must strive to assess these other health factors to improve the utility of genetic data in diverse patient populations. Partnerships among patients and providers are needed to better educate patients on the value of genetics in health care, and reassuring patients that past misconduct will not be repeated [56]. To this end, our providers are working to increase minority participation in genetics and genomics research as we are a recruitment center for The AoURP and for studies by collaborators in the Weitzman Institute (the first community-based research center established by an FQHC). Thus, provider education in genetics and genomics not only can help facilitate integration into clinical practice but promote research opportunities to patients and prepare providers to respond to patient inquiries and review of results from the AoURP.

There are some limitations to this study to be noted. The survey was administered at a single FQHC and the views may not be representative of other providers practicing in FQHCs or providing care for minority patients. However, the integration of genetic services in primary care at FQHCs is relatively new [32], and many challenges exist, including provider knowledge [57]. Our participation in the AoURP provides an opportunity for both our providers and patients to increase their awareness of genomics and gain invaluable experience in discussing results with patients. As we move forward in our development of a genomic medicine program, we now have a better understanding of the challenges. We will focus our efforts in these key areas to identify solutions. Participation may have been biased toward those with interests in or positive attitudes about genetics and genomics. Future studies are needed to explore the acceptability for service delivery through the referral process, including specific genetic tests and counseling services. Additionally, an understanding of patients' attitudes, needs, and barriers around genomic medicine services will be an important component to fully developing and implementing a genomic medicine program.

In conclusion, while the providers at our FQHC show many similarities between attitudes and perceived knowledge to previously published studies [11], these findings can be used to prepare providers for bringing genomic medicine to underserved populations. Our providers believe that patients will be interested in genetic services but recognize their limited knowledge and capabilities. As community health centers may be good settings to conduct pragmatic clinical studies on validation and delivery of genetic applications with populations historically underrepresented in biomedical and clinical research, providers at these locations will need to have access to educational resources to

help recruit patients and address research questions. Integration of genomic medicine in community health centers will be critical for mitigating health disparities faced by ethnic and racial minorities. Community health centers are only just beginning to develop genomic medicine programs [58], and will need to tackle many challenges, including accountability, monitoring, and information sharing. A first step is ensuring that providers have adequate training and resources to appropriately deliver genomic-based care. The results of this survey further validates the continued need to develop genetics and genomics training and support for primary care providers, while highlighting that additional staff and patient support is needed, in order to provide patients with access to new genetics and genomic services and minimize health disparities as well as participating in research opportunities.

Statement of Ethics

Community Health Center, Inc. Institutional Review Board (IRB) approved this study. The study ID is IRB #1155. All subjects gave informed consent prior to participating in this study. A waiver of written consent was granted by the IRB because no identifiable information was collected and the study posed minimal risks.

Conflict of Interest Statement

The authors have no relevant financial or nonfinancial interests to disclose.

Funding Sources

No funding was received for conducting this study.

Author Contributions

Yashoda Sharma and Veena Channamsetty contributed to the study conception and design. Material preparation, data collection, and analysis were performed by Yashoda Sharma, Livia Cox, Lucie Kruger, and Susanne B. Haga. The first draft of the manuscript was written by Yashoda Sharma, Susanne B. Haga, and Veena Channamsetty, and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

Data Availability Statement

All data generated or analyzed during this study are included in this article. Further inquiries can be directed to the corresponding author.

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