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Integrating Genomic Screening into Primary Care: Provider Experiences Caring for Latino Patients at a Community-Based Health Center

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Abstract

Introduction: Minority communities have had limited access to advances in genomic medicine. Mayo Clinic and Mountain Park Health Center, a Federally Qualified Health Center in Phoenix, Arizona, partnered to assess the feasibility of offering genomic screening to Latino patients receiving care at a community-based health center. We examined primary care provider (PCP) experiences reporting genomic screening results and integrating those results into patient care. **Methods:** We conducted open-ended, semi-structured interviews with PCPs and other members of the health care team charged with supporting patients who received positive genomic screening results. Interviews were recorded, transcribed, and analyzed thematically. **Results:** Of the 500 patients who pursued genomic screening, 10 received results indicating a genetic variant that warranted clinical management. PCPs felt genomic screening was valuable to patients and their families, and that genomic research should strive to include underrepresented minorities. Providers identified multiple challenges integrating genomic sequencing into patient care, including difficulties maintaining patient contact over time; arranging follow-up medical care; and managing results in an environment with limited genetics expertise. Providers also reflected on the ethics of offering genomic sequencing to patients who may not be able to pursue diagnostic testing or follow-up care due to financial constraints. **Conclusions:** Our results highlight the potential benefits and challenges of bringing advances in precision medicine to community-based health centers serving under-resourced populations. By proactively considering patient support needs, and identifying financial assistance programs and patient-referral mechanisms to support patients who may need specialized medical care, PCPs and other health care providers can help to ensure that precision medicine lives up to its full potential as a tool for improving patient care.

Keywords

genomic screening, individualized medicine, primary care, health disparities, federally qualified health center

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Introduction

There has been significant interest in precision medicine as a tool to understand disease and optimize patient care at an individual level.[1] To date, however, the majority of precision medicine initiatives have been positioned in academic medical centers and large healthcare systems.[2,3] As a result, the potential health benefits of integrating new forms of precision medicine into community-based health centers that provide primary care services to more diverse communities are unclear.[4,5]

Integrating genomic medicine into community-based health centers presents several challenges. Many primary care providers (PCPs) report a lack of familiarity with genetic testing and genomic screening.[6-8] Absent genetic counseling resources, PCPs working in community-based health centers may find it difficult to educate patients about

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genetic testing options, establish clinical management plans informed by genetic test results, or obtain insurance coverage for their patients.[9,10] Additionally, patients from lower-resource communities, and the physicians who care for them, are concerned about the affordability of genomic medicine and its potential to widen existing health disparities.[11-13]

To better understand the potential value and challenges of integrating genomic medicine into community-based health centers, we established a partnership between Mayo Clinic and Mountain Park Health Center (MPHC), a Federally Qualified Health Center in Phoenix, Arizona that provides primary care and behavioral health services to financially disadvantaged patients. We offered genomic screening to interested patients, incorporated those results into patients' electronic health record (EHR), and assessed the impact of genomic screening on patients and healthcare providers.[14,15]

We describe this partnership in greater detail, focusing on the experiences of PCPs and clinical staff who supported this genomic screening initiative and counseled patients who received positive genomic screening results. Examining both the potential benefits and burdens of integrating genomic screening into community-based health centers that provide care to lower-income patients can help to ensure that precision medicine lives up to its full potential as a tool for improving patient care.

Methods

We invited 1621 patients at MPHC to undergo genomic screening. These patients self-identified as Latino and all had previously provided a blood sample to the Sangre Por Salud Biobank.[15] Of those invited, 500 patients agreed to participate after attending an in-person genetics education session and informed consent discussion, which was conducted in either Spanish or English depending on participant preference. Participants agreed to have their biobank sample analyzed, receive genomic screening results, and have those results placed in their EHR.[16]

Genomic analysis included the sequencing of 68 genes known to be associated with disease and screening for 14 actionable single nucleotide variants.[14] After genomic analysis was completed, we contacted participants whose results revealed a pathogenic or likely pathogenic (P/LP) genetic variant by certified mail or telephone and asked them to schedule an in-person appointment. At this appointment, a medical geneticist who was a member of the research team disclosed the patient's genomic screening results and discussed their potential health implications. An interpreter participated in these discussions when needed. Following this appointment, genomic screening results were placed in the patient's EHR and an alert was sent to the patient's PCP. Participants whose results indicated no P/LP variants were notified about their results by mail, with subsequent confirmation of receipt by a study staff member and optional in-person support available by request.

PCPs were encouraged to discuss their patient's genomic screening results with the medical geneticist who had met with these patients. All PCPs who received an alert about a patient with a positive screening result elected to consult with the medical geneticist in-person. During these one-on-one consultations, PCPs received individualized genomic education focused on the specific results reported to their patient(s). These consultations provided PCPs with an opportunity to discuss the mechanism of disease pathology, the penetrance of the genetic mutation, the need for medical surveillance, and potential clinical management options. The medical geneticist also informed each of the PCPs about diagnostic criteria and clinical practice guidelines relevant to their patients' results.

To assess provider experiences caring for patients with P/LP results, we conducted semi-structured interviews. These interviews were conducted in-person or by telephone, depending on provider availability, approximately 3 months after their consultation with the medical geneticist and subsequent interactions with their patients. To provide a comprehensive assessment of these experiences, we interviewed PCPs, the medical geneticist, and the primary clinical research coordinator at MPHC who coordinated the reporting of genomic results and supported clinical staff involved in the care of patients who received a P/LP result. The medical geneticist (N.M.L.) and research coordinator (V.H.) were members of the study team and are co-authors of this report. We included them as participants in this study given their critical roles in patient care and provider support, and to capture their insights into potential operational challenges associated with providing genomic screening in a community-based health center.

Interviews were conducted by 2 experienced qualitative researchers (R.R.S., E.J.S.), who asked providers to comment on their experiences caring for recipients of P/LP results. Interviewees were also asked to reflect more broadly on their perceptions of the potential benefits and challenges of integrating precision medicine into their clinical practice. All interviews were audio-recorded and transcribed verbatim by a professional transcription service. Two members of the research team (T.S., E.J.S.) read the transcripts and conducted a descriptive, thematic analysis. The first author wrote detailed thematic memos, which were reviewed and revised iteratively by the analytic team (T.S., E.J.S., R.R.S.).

Results

Genomic screening was provided to 500 patients at MPHC. Of these individuals, ten were found to have a P/LP result.[17] Table 1 highlights the diversity of clinical management

Table 1. Illustrative Cases of Genomic Screening Results and Their Management at a Community Health Center.

Patient history	Medical risks and management	Outcomes
A 46-year-old female patient screened positive for a *BRCA1* variant, indicating potential hereditary breast-ovarian syndrome and a high risk of cancer. This patient was uninsured and had phased out of the MPHC system, not having seen a provider there in approximately three years. The patient did not report any significant family history of cancer. She was primarily Spanish-speaking and had <9th grade education, which posed some concerns about comprehension in receiving the genetic result.	Risk of cancer over next 10 years • Breast cancer 25% • Ovarian cancer 10% Lifetime risk of cancer • Breast cancer 46-87% • Ovarian cancer 39-63%[a] ***Recommended Management*** Recommendations (for women): Clinical breast exam every 6-12 months Breast imaging annually Recommend salpingo-oophorectomy Consider mastectomy Share results with male and female relatives[b]	Due to this patient's extended time outside of the MPHC system, it took several attempts at contact before the study team was able to bring the patient in for a consultation. Despite the lack of a family history of cancer, this *BRCA1* variant was still deemed pathogenic. Given the patient's financial situation, the provider felt that discussing prophylactic surgery would be inappropriate at this time. The provider gave instructions for monthly self-breast exams, emphasizing the importance of these in the absence of frequent clinical breast exams. The provider limited recommended management to yearly mammography in the hopes that the patient would follow up. The provider is open to the possibility of discussing more intensive options should the patient's insurance status change.
A 48-year-old male patient screened positive for a *BRCA2* variant, indicating potential hereditary breast-ovarian syndrome and a high risk of cancer. This patient had 6 relatives with a history of cancer; 2 originated in the breast, 1 in the colon, and 3 with an unknown primary site. The ages of onset for these individuals were unclear. He was primarily Spanish-speaking and had some high school education, necessitating the use of an interpreter.	Lifetime risk of cancer • Breast cancer (male) up to 8.9% • Prostate cancer - 20% • Pancreatic cancer 2-7%[a] ***Recommended Management*** Recommendations (for men): Breast self-exam Clinical breast exam annually Regular prostate cancer screening Smoking cessation (if necessary) Share results with male and female relatives[b]	This *BRCA2* variant was deemed pathogenic due to the strong family history of cancer. The patient initially met with the medical geneticist to discuss the positive result, followed by a consultation with the provider on the same day. The patient appeared concerned when meeting with the medical geneticist, but appeared relatively at ease by the time he met with his provider. The provider went over self-breast exam procedures and instilled the importance of discussing the results with family members. Information on female risks was also given. The patient followed up within a month after reporting "feeling a lump" in his breast; this lump was not unusual and deemed not of clinical concern. During this follow-up visit, a consultation with behavioral health indicated that the patient appeared rather nervous and had been checking himself quite frequently. The provider assuaged the patient's concerns and recommended limiting self-exams to monthly frequency. The provider also confirmed that the patient had discussed the genetic results with his sisters, and that he had urged them to also pursue *BRCA* screening.
A 50-year-old female patient screened positive for a *FBN1* variant, indicating a risk for cardiovascular and connective tissue disorders, including Marfan syndrome. The patient reported some connective tissue pathologies in her history. These included congenital dislocation of the lenses (an *FBN1*/Marfan feature) and early bilateral hip replacement in her 40s. During these hip replacements, the patient's heart was found to be enlarged and there was a small dilatation of the aorta. The patient was obese and hypertensive and body build was not typically Marfanoid. The patient's family exhibited aortic pathology. Her father had died young of an aortic rupture. Her sister had undergone an aortic replacement. The patient's brother, who had been "tall and thin," died at age 25 of unknown cause.	Cardiovascular risks in Marfan Syndrome • Progressive aortic dilatation • Risk of dissection/rupture by age 60-60% • Valve dysfunction Ocular risks • Progressive myopia • Risk of lens displacement - 60% • Retinal detachment, glaucoma, cataracts Other risks • Scoliosis • Tendency for hernias • Spontaneous pneumothorax • Dural ectasia[c] ***Recommended Management*** Evaluation by clinician familiar with Marfan syndrome	The procedure of handling this positive result differed from the provider's standard management of Marfan-type findings. The variant was deemed likely pathogenic for some cardiovascular or connective tissue conditions, and the patient and family history indicated the need for a Marfan workup. The patient had only a small dilatation of the aorta, raising some uncertainty as to appropriate, cost-effective management given the patient's Medicaid status. The provider emphasized the importance of following up regularly with her cardiologist; they also gave lifestyle advice to lower her weight and blood pressure. The provider felt that it was necessary to discuss the results and further screening with the patient's sons and sister, the latter having had a strong phenotypic presentation of cardiovascular dysfunction. The sister was in the MPHC system; the study team had re-established contact, but they had not been able to bring her in for a consultation yet. Neither the patient nor the study team had been able to make progress in discussing further actions regarding the genetic results with the patient's sons. These individuals were employed in construction jobs with heavy physical labor; the patient expressed concern that diagnosis of a cardiovascular condition might render her sons unable to continue work or obtain insurance. Thus, the provider began exploring options to forward the result to the patient's sons' providers, in hopes that the sons might follow up with their PCP's.

(continued)

Table 1. (continued)

Patient history	Medical risks and management	Outcomes
	Baseline echocardiogram with measure of aortic root and ascending aorta annually • Consider CT or MRI of vasculature • Evaluation for valvular insufficiency • Initiate beta blocker or Angiotensin II receptor blocker for cardioprotection • SBE prophylaxis if mitral or aortic regurgitation • If 5.0 cm aortic root, involve CT surgeon Ophthalmologic evaluation annually Avoidance of contact sports, isometric exercises, cardiovascular stimulants, vasoconstrictors, LASIK surgery, breathing against resistance[c]	
A 53-year-old woman screened positive for a *PALB2* variant, which may confer increased susceptibility to breast and pancreatic cancer. Due to the patient's limited health literacy and not being in contact with relatives, it was difficult for providers to attain a reliable history beyond first-degree relatives. The patient did not report any immediate family members with a history of cancer. The patient was uninsured and had transitioned outside of the MPHC system by the time the results were returned.	Risk of breast cancer by age 70 up to 35%[d] Potential increased risk for pancreatic cancer[e] ***Recommended Management*** Breast imaging annually Clinical breast exam every 6-12 months Mastectomy can be discussed if strong family history Pancreatic cancer screening if first-degree relative with pancreatic cancer Smoking cessation (if necessary)[f]	This *PALB2* variant was deemed pathogenic. Clinical recommendations, including pancreatic imaging, hinge upon having a family history of pancreatic cancer; however, this was unknowable given the sparse family history. The patient was advised to continue yearly mammograms, but few other screening recommendations or referrals could be carried out in the absence of stronger clinical evidence. There were concerns that the patient might be burdened with out-of-pocket costs without warrant if more frequent or costly measures were recommended.
A 37-year-old woman screened positive for a *TP53* variant, which is typically associated with Li-Fraumeni syndrome, a hereditary predisposition to cancer. The provider was able to obtain an oral family history from the patient, who reported a large family, including 7 children and 15 nieces and nephews. Other than the patient's mother, who had uterine cancer in her late 40s, the patient reported no other cancer in the family history. The patient was Spanish-speaking and uninsured.	Elevated lifetime risk of cancer up to 70% (men) or 90% (women) Examples of LFS-associated cancers: • Adrenocortical carcinomas • Breast cancer • CNS tumors • Osteosarcomas • Soft-tissue sarcomas[g] ***Recommended Management*** Physical exam (with skin & neuro) annually Breast MRI annually starting age 20 (add mammography age 30) Avoidance of unnecessary radiation Consider whole body and brain MRI annually Colonoscopy & UGI endoscopy every 2-5 years In children, biochemical screening for adrenal tumors[h]	The patient met with the medical geneticist to receive the result of this pathogenic variant of *TP53*. However, follow-up appointments with the provider to discuss clinical management were not scheduled on the same day. The patient did not arrive for the appointment with the provider; thus, little could be conveyed to the patient regarding future screening and referrals. Given the limited family history of cancer, the provider would have recommended regular physical exams and cancer screening for the patient. Discussion of cascade screening for immediate family members would have also been appropriate.

(continued)

Table 1. (continued)

Patient history	Medical risks and management	Outcomes
A 24-year-old woman screened positive for a variant of *SCN5A*, a gene associated with a range of cardiac arrhythmias (especially ST and QT abnormalities). The patient was on Medicaid and provided a patient and family health history. The patient did not report any contributory health issues in the extended family history, including eight adult first-degree relatives. The patient herself was asymptomatic and did not have any major pre-existing conditions.	Variety of cardiac rhythm abnormalities • Brugada syndrome (ST abnormalities with risk of ventricular arrhythmias) • Long QT syndrome (tachyarrhythmias) • Sick sinus syndrome • Multifocal ectopic Purkinje-related premature contractions • Isolated cardiac conduction defect • Atrial fibrillation[h] May cause unexplained fainting, seizing, recurrent palpitations Sudden death (mean age 40 years) and SIDS[i] ***Recommended Management*** Obtain targeted family history and ECG Referral to cardiac electrophysiologist Discussed that for symptomatic Brugada syndrome, consider quinidine daily[i] For Long QT syndrome, beta blocker therapy (even if asymptomatic)[j] Avoidance of swimming, sudden startles, stimulants, anesthetics, electrolyte disturbances Consider implantable defibrillator[j]	The patient met with the medical geneticist to discuss this likely pathogenic variant of *SCN5A*. The study coordinators reported some difficulty establishing contact with this patient, a young woman who had not frequently obtained care at MPHC. The patient was scheduled to follow up with her provider on a separate day to discuss clinical management of the condition, but she did not appear for this consultation. The provider expressed concern that the patient might have been overwhelmed by this genetic result, due to its association with sudden death syndrome, an association readily made on internet searches. The provider also expressed concern with the inability of the study coordinators to establish further contact given the PCP's potential liability if the patient were to be affected by a sudden cardiac event, especially if cardiac evaluation and follow-up management was not established.

[a]Petrucelli N, Daly MB, Pal T. BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer. 1998 Sep 4 [Updated 2016 Dec 15]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020.

[b]National Comprehensive Cancer Network. Genetic/Familial High-Risk Assessment: Breast and Ovarian (Version 3.2019). https://www2.tri-kobe.org/nccn/guideline/gynecological/english/genetic_familial.pdf. Accessed April 27, 2020.

[c]Dietz H. Marfan Syndrome. 2001 Apr 18 [Updated 2017 Oct 12]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020.

[d]Antoniou AC, Casadei S, Heikinnen T, et al. Breast-Cancer Risk in Families with Mutations in PALB2. *NEJM*. 2014 Aug 7; 371: 497-506.

[e]National Comprehensive Cancer Network. Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic (Version 1.2020). https://jnccn.org/view/journals/jnccn/18/4/article-p380.xml. Accessed April 27, 2020.

[f]National Comprehensive Cancer Network. Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic (Version 1.2020). https://jnccn.org/view/journals/jnccn/18/4/article-p380.xml. Accessed April 27, 2020.

[g]Schneider K, Zelley K, Nichols KE, et al. Li-Fraumeni Syndrome. 1999 Jan 19 [Updated 2019 Nov 21]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020.

[h]Wilde AAM, Amin AS. Clinical Spectrum of SCN5A Mutations. *JACC: Clinical Electrophysiology*. 2018 May; 4(5): 569-579.

[i]Brugada R, Campuzano O, Sarquella-Brugada G, et al. Brugada Syndrome. 2005 Mar 31 [Updated 2016 Nov 17]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020.

[j]Alders M, Bikker H, Christiaans I. Long QT Syndrome. 2003 Feb 20 [Updated 2018 Feb 8]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle, WA: University of Washington, Seattle; 1993-2020.

scenarios associated with reporting medically actionable genomic screening results and provides a summary of select patient health histories, genomic results, clinical management options, and recommended follow-up care. These cases illustrate the types of care-coordination challenges that PCPs may encounter when genomic screening is offered at a community-based health care center like MPHC.

The medical geneticist returned genomic screening results to 9 of the 10 patients with P/LP results (One participant was scheduled to receive screening results, but presented for their appointment in severe psychological distress due to unrelated personal circumstances. Given these circumstances, the health care team decided that the disclosure of genomic screening results should be postponed. However, subsequent attempts to return those results have not been successful). Immediately following their appointment with the medical geneticist, patients met with a behavioral health provider to assess their psychosocial needs and support. At that time, an appointment was scheduled with a PCP to discuss clinical management and follow-up care. Seven of the nine patients who received P/LP results attended their appointment with a PCP.

At the time we scheduled interviews to discuss provider experiences, one of the MPHC PCPs had retired and another had left MPHC. We were able to interview all of the remaining five PCPs, the medical geneticist, and the primary clinical research coordinator for a total of seven participants. Interviews ranged in length from 15 to 120 minutes.

Benefits of Genomic Screening

All interviewees felt that implementing genomic sequencing at MPHC had benefited patients, providers, and the community at large. They emphasized the value of genomic screening to identify unknown risk factors. Several providers also mentioned that screening for familial susceptibility to cancer or heart conditions can raise awareness of the importance of regular medical monitoring and engagement with healthcare providers. Providers noted that by implementing screening for younger, asymptomatic patients, providers might be able to contextualize patients' health behaviors in terms of personalized risk. They also noted that those screened might influence the behavior of their at-risk family members and others in the community. Providers often remarked that genomic screening was closely aligned with the goals of primary care, which include proactive health monitoring and the cultivation of positive health behaviors: *"[The patients], they wanna take a proactive measure and know, 'Hey, do I have a predisposition to some kind of genetic disorder, and could my family benefit from knowing this information?'" (ID5)*

Other benefits noted by interviewees included the perception that genomic screening could serve as an educational tool to empower individual patients and their families to learn more about disease risks. Specifically, interviewees felt genomic screening could be useful in discussing family health histories and addressing knowledge gaps regarding disease histories: *"People don't actually know their family history beyond their first-degree relatives for the most part. They may have died of cancer in Mexico, but Lord knows what it was. You can't get records" (ID7).* Several providers also remarked that genomic screening could help improve health literacy for patients and their families. Interviewees felt that patients who pursued genomic screening might be better positioned to engage with clinical information and understand the interplay of genetic and lifestyle factors on their health outcomes: *"[Even] if they do nothing, no action, they at least know a little bit more about genetics" (ID6)*.

The perceived benefits of genomic screening were not limited to patients and their families. Some providers expressed a personal desire to remain at the cutting-edge of primary care medicine and viewed their involvement in translational research as critical to that end: *"If you don't get involved with research, you kind of get left behind" (ID7).* Other providers viewed learning more about precision medicine as critical to their practice: *"That's where medicine is going if we like it or not" (ID4).*

Given the breadth of genomic screening, and the diversity of potential patient management scenarios, PCPs greatly appreciated the tailored genomics education they received for their patients from the medical geneticist supporting the genomic screening initiative. Several providers acknowledged a lack of familiarity with genomic screening methods and the genes evaluated in the study: *"I wasn't really aware of how far or how, um, advanced some of these tests and interpretations has gotten, really" (ID1)*. They described the clinical decision support that the medical geneticist provided as invaluable, particularly in relation to advice on clinical management and medical monitoring plans.

Challenges Encountered in Offering Genomic Screening

Providers described several struggles and frustrations that they encountered, particularly related to reporting genomic results and coordinating follow-up care. A common challenge was difficulty contacting patients and conveying a sense of urgency to come into clinic to discuss their results and arrange for follow-up care: *"She never answered the phone. She never responded to the letter" (ID6).* Providers reported similar challenges contacting at-risk family members and encouraging them to come in to discuss genetic testing options.

Several providers voiced concerns about their patients' capacity to pursue recommended follow-up care due to financial constraints. This was noted as a source of

considerable personal distress to the PCPs. Since many patients who receive care at MPHC are underinsured, providers worried that patients with P/LP variants would not have sufficient insurance to cover the costs of follow-up care. As a result, those patients might be left with a difficult decision to either pay out-of-pocket expenses or forego recommended diagnostic evaluations or procedures: *"Most of these patients can hardly afford their blood pressure medicine so I don't expect them to afford expensive procedures" (ID4).* Providers described inadequate patient health insurance and a lack of subsidized government alternatives as significant barriers to patients accessing the care they need, as defined by clinical practice guidelines for the management of P/LP results.

This concern about patients' inability to act on medical recommendations based on genomic results prompted several PCPs to question whether genomic screening should be offered to individuals who do not have the financial capacity to pursue follow-up care in the event of a positive result: *"I have to be realistic. I mean, if we're not gonna pay for it, we shouldn't be ordering it" (ID3).* This tension was experienced as a form of moral distress for some providers, exacerbated in instances when a patient or an at-risk family member did not appear for their follow-up clinical visit: *"I wonder what my legal responsibility is if she hasn't come back. I'd probably need to look her up and send her a certified letter to make sure she comes in. I was worried about her" (ID1).*

Suggestions for Future Genomic Screening Initiatives

While opinions about the clinical impact of precision medicine varied among PCPs, with some voicing excitement and others apprehension, all of the providers we interviewed expected that their patients would have more questions related to genetic testing and genomic screening in the future. As providers reflected on their experiences counseling patients about genomic results, they had several suggestions to support future efforts to bring genomic screening to community-based health centers. Text Box 1 describes these recommendations, many of which were related to anticipating patient-support needs, including financial costs associated with additional diagnostic tests, and coordinating specialized medical care that might require referral to another healthcare facility.

Text Box 1. Provider or Interviewee Recommendations for Offering Genomic Screening in a Community-Based Health Center.

Ensure pre-test counseling is available in the patient's language of preference
Have a medical geneticist or genetic counselor available on-site to help providers interpret genomic test results and develop care-management plans
If possible, involve a case manager to assist patients in sharing screening results with at-risk family members and to maintain patient engagement over time
Identify financial assistance mechanisms to support underinsured patients who may need confirmatory diagnostic tests or specialized medical care
Prioritize genomic screening services that impact ongoing patient-care activities, such as pharmacogenomic screening related to medications that are frequently prescribed in community-based health centers

Discussion

Our findings highlight tensions in bringing precision medicine to community-based health centers. On the one hand, PCPs wanted to ensure that individuals from lower-resource communities are part of the research driving the future of medicine, in part to ensure that their patients are able to benefit from those advances. On the other hand, financial constraints contributed to a number of clinical management challenges, resulting in moral distress and prompting some providers to ask whether it was ethical to offer genomic services in lower-resource settings.

Although providers noted the potential of genomic screening to provide their patients with clinical and preventive health benefits, they found it difficult to get patients and their family members to come in for primary care consultations and follow-up appointments. Additionally, many of the patients seen in the clinic lacked adequate health insurance coverage and the financial resources to pursue advanced diagnostic evaluations and specialized care available at referral facilities. These challenges are often faced by PCPs and other clinicians who practice in community-based health centers serving low-income populations, contributing to physician burnout and dissatisfaction.[18-20] These and other burdens on providers are important to consider as new forms of precision medicine are integrated into primary-care clinics.

Even with the additional personnel and specialist support available through the collaboration with Mayo Clinic, PCPs experienced moral distress caring for patients who received medically actionable genomic screening results. The concept of moral distress has garnered considerable interest in the medical community, resulting in a growing body of scholarship examining moral distress resulting from the care of uninsured patients whose healthcare needs are not being adequately met.[21-25] In our study, providers

highlighted the challenge of getting uninsured and underinsured patients at high risk of disease the medical care they felt was necessary given their genomic results and cited concerns about the lack of state or federal funding to assist patients with insufficient financial resources to pay for recommended medical care. These challenges, combined with difficulties getting patients and family members to attend follow-up care appointments, were noted as significant contributors to providers' moral distress.

Consistent with our findings, prior studies have underscored the difficulty of arranging financial coverage for cascade genetic testing (testing of at-risk family members of the proband after initial return of a P/LP variant).[26,27] Addressing these and other health inequities in genetic medicine requires that we consider how best to make the potential benefits of precision medicine available to under-resourced communities.[11,28,29] This sentiment was evident in our interactions with PCPs, all of whom expressed a strong interest in advancing community-based health by promoting genomic research, despite limited evidence of clinical utility in comparable settings and full awareness of the many challenges associated with the clinical management of patients who received a medically actionable result. A consistent sentiment among PCPs was enthusiasm for this research in genomic screening to learn more and consider how precision medicine might benefit their patients in the future.

Our findings also underscore a need for system-wide provider education and clinical decision support as a key element of integrating genomic screening into community-based health centers. Even when the medical implications of genetic test results are reviewed directly with patients (as they were in our study), future providers will have access to this information via the electronic health record and will need to be prepared to integrate those genetic test results into ongoing patient care activities. All of the PCPs we interviewed expressed a lack of familiarity with clinical genetic testing and the management of positive genomic screening results prior to this study, which is consistent with the broader literature.[6-8] Similarly, other genetic implementation studies in primary care settings have demonstrated a clear need for ancillary physician education, often through partnerships with academic medical centers.[30-32] These academic partnerships can also help to address infrastructure limitations, for example, by providing referral options for complex patients who would benefit from additional evaluation.[33-35] While productive, this reliance on outside academic institutions raises questions about the long-term sustainability of genomic screening as a service provided by community-based health centers. Empowering on-site PCPs through genomic education and clinical decision support integrated into the EHR may provide more stable long-term support for the integration of precision medicine into community-based health centers.[35,36]

Lastly, our findings suggest that precision medicine may not integrate seamlessly into community-based health centers that support medically under-resourced communities. As advocates of precision medicine seek to expand its reach to include underrepresented populations in biomedical research, it will be critical that they evaluate experiences at community-based health centers. While there are many potential benefits of incorporating genomic screening into primary care, the burdens on patients and their PCPs may be considerable in lower-resource settings.[37]

It is important to note the limitations of our results. This study examined the experiences of PCPs caring for Latino patients at a single community-based health center serving a primarily low-income patient population. Although we interviewed all of the available PCPs at MPHC who cared for a patient with an actionable genomic screening result, the experiences of these providers may not be typical of PCPs at other facilities or in other communities. Additionally, since 2 of the 9 patients had limited healthcare interactions after receiving their genomic results, the experiences of their PCPs may not be typical of other providers caring for patients with medically actionable screening results.

Despite these limitations, our findings help to address a significant gap in available scholarship by describing provider perspectives on the integration of genomic screening into community-based health centers, a setting in which patients are rarely offered new forms of precision medicine despite the potential for genomic screening to improve their health through the identification of unknown, but medically manageable risk factors.

Conclusion

Avoiding potential inequities that might result from advances in precision medicine will require creative approaches to delivering genomic services. By examining the potential benefits and challenges of offering genomic screening in community-based health centers, particularly health centers that support lower-income patients from diverse racial and ethnic backgrounds, we can transform what might otherwise be a highly disruptive and potentially discriminatory technology into a useful, positive influence on patients.

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