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Are Physicians Prepared for Whole Genome Sequencing? A Qualitative Analysis

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Abstract

Background—Although the integration of whole genome sequencing (WGS) into standard medical practice is rapidly becoming feasible, physicians may be unprepared to use it.

Methods—Primary care physicians (PCPs) and cardiologists enrolled in a randomized clinical trial of WGS received genomics education before completing semi-structured interviews. Themes about preparedness were identified in transcripts through team-based consensus-coding.

Results—Data from eleven PCPs and nine cardiologists suggested that physicians enrolled in the trial primarily to prepare themselves for widespread use of WGS in the future. PCPs were concerned about their general genomic knowledge, while cardiologists were concerned about how to interpret specific types of results and secondary findings. Both cohorts anticipated preparing extensively before disclosing results to patients by using educational resources with which they were already familiar, and both cohorts anticipated making referrals to genetics specialists as needed. A lack of laboratory guidance, time pressures, and a lack of standards contributed to feeling unprepared.

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CONFLICT OF INTEREST

The authors report no conflicts related to the work presented herein.

Discussion—Physicians had specialty-specific concerns about their preparedness to use WGS. Findings identify specific policy changes that could help physicians feel more prepared, and highlight how providers of all types will need to become familiar with interpreting WGS results.

Clinical Trials Registration—ClinicalTrials.gov # NCT01736566.

Keywords

whole genome sequencing; physicians; preparedness; self-efficacy; genetic literacy; qualitative research

INTRODUCTION

Whole genome sequencing (WGS) may soon play an important role in primary and specialty care. Falling technical costs and turnaround time are making widespread WGS use feasible (1–4), and it is already useful for diagnosing disease, informing treatment decisions, and assisting life decisions (1, 5–8). Genomic sequencing is a key component of President Obama's Precision Medicine Initiative (9), and healthcare systems are developing the infrastructure to enhance the utility of genomics (10, 11). The era of genomic medicine is nearing.

Many physicians may be reluctant to engage in it, however, because they feel unprepared. WGS results are far more complex than other medical or genetic tests. Surveys have found that physicians of all types often lack genomic literacy (12–15) and frequently feel unprepared to use or respond to even single gene testing, especially primary care physicians (12, 16–18). Medical training and continuing education programs have expanded to better address genomics (19), but the amount of training that is specific to sequencing is limited.

Understanding how prepared physicians feel to use WGS can provide valuable insight about how to tailor educational programs and develop infrastructure to support genomic medicine. Contrasting the perspectives of primary care physicians (PCPs) and specialists may be particularly valuable. Specialists, including cardiologists, are likely to practice *disease-specific genomic medicine* where the genome is interrogated to identify causes for particular presentations, such as a strong family history of cardiomyopathy. In contrast, PCPs are likely to practice *general genomic medicine* where the genome is examined as part of routine preventive medicine to identify risks of future disease and to assist decision making (20).

Here, we summarize interviews with physician participants of a randomized trial of WGS. We first describe how prepared they felt to use WGS, comparing the perspectives of PCPs and cardiologists. We then describe expectations about developing competencies, and identify factors that made physicians feel prepared or unprepared about WGS. Findings inform recommendations to enhance physicians' abilities to integrate sequencing into their practices.

MATERIALS AND METHODS

Overview and Participants

We report on data from the MedSeq Project, a randomized trial of WGS in clinical care. Study procedures are published in detail elsewhere (20). Briefly, we enrolled PCPs and cardiologists from a large urban network of academic hospitals and outpatient practices. To recruit PCPs, JLV introduced the study at staff meetings of seven group practices and sent emails to individual PCPs. To recruit cardiologists, study investigators and enrolled cardiologists approached colleagues. Physicians provided informed consent at the first group education session. After enrollment, they completed a "just in time" educational curriculum consisting of two 1-hour in-person group sessions taught by medical geneticists and genetic counselors. Sessions focused on the following content: an overview of genomic sequencing; contextualizing WGS results with other health information; Mendelian inheritance patterns; genomic support resources; risk prediction; and MedSeq Project WGS reports. Physicians also completed 12 self-paced online modules, designed to take 4 hours to complete (see online Supporting Information). They received six hours of continuing medical education credits and financial incentives for participating.

As the intervention of interest, physicians received WGS reports (21) for a sample of their patients. Physicians viewed example reports during the in-person education sessions, and they learned that they could seek assistance from a Genome Resource Center (GRC) consisting of genetic counselors and medical geneticists (see Table 1). The study team also created an online repository for educational materials.

The MedSeq Project protocol was developed by a multidisciplinary team with expertise in laboratory and clinical genetics, bioinformatics, health economics, health behavior and health policy. The Partners Human Research Committee and Baylor College of Medicine Institutional Review Board approved the study protocol.

Data Collection and Analysis

Physicians provided demographic and practice information on self-administered questionnaires. They also reported *genetics training* "beyond the typical medical school curriculum" and *frequency discussing genetics* with patients. Before and after education, we assessed *knowledge* using six multiple-choice items developed by the study team; *perceived preparedness* by asking, "How prepared do you feel about disclosing results [from this study] directly to your patients?"; and *self-efficacy* about genetic testing using a 5-item scale (22). Missing post-education data were imputed from pre-education surveys. We used Fisher's exact tests and t-tests to compare characteristics of PCPs and cardiologists.

Semi-structured interviews, about 45 minutes long, were conducted after education, but before disclosure visits with patients. An interview guide (see online Supporting Information) facilitated discussions about motivations for study participation, experiences with genetic testing, and attitudes about WGS. During interviews, physicians viewed WGS reports and discussed potential benefits and concerns; expectations about disclosing sequencing results; and beliefs about the impact of WGS on clinical care.

The study team identified themes from interview transcripts following standard procedures for team-based qualitative analysis and consensus-coding (23–25). Analyses focused on differences between PCPs and cardiologists on themes related to physicians' preparedness to disclose and respond to WGS reports. We coded a set of transcripts using inductive methods to identify recurring themes (26). We then re-coded all transcripts using a formalized list of themes. Interview data were managed using ATLAS.ti version 7.

RESULTS

Sample characteristics

Of 10 cardiologists and 90 PCPs approached to participate, nine cardiologists (90%) and eleven PCPs (12%) enrolled (see Table 2). Non-participating physicians cited a lack of time and concern about clinical workflow interruption. One enrolled PCP reported additional genetics training via a "genetic course for PCPs", while four cardiologists reported additional training via basic science research (C02), PhD training (C08), postdoctoral research (C03), and continuing medical education (C06). Five of nine (56%) cardiologists reported discussing genetic information with patients "almost always" or "often" compared to one of eleven (9%) PCPs (p=0.050). Five of nine cardiologists (56%) also reported feeling prepared or very prepared to disclose results, compared to two of 11 (18%) PCPs (p=0.127). Cohort differences were not observed on mean scores for knowledge (PCPs: 5.0 of 6 items correct, cardiologists: 5.4, p=0.266) or self-efficacy (PCPs: 3.5 on 1–5 scale, cardiologists: 3.2, p=0.336), although education improved self-efficacy more for PCPs than cardiologists (Δ=+0.4 vs −0.1, p=0.034). One cardiologist and one PCP were not interviewed.

Three primary themes related to preparedness emerged during analysis of transcripts: *genetic literacy*, *concerns about preparedness*, and *motivations about developing proficiencies*. Table 3 summarizes these and key secondary themes.

Genetic Literacy and Preparedness

Physician interviews reflected both confidence and apprehension. Both cohorts discussed WGS as a shift away from traditional genetic testing. PCPs tended to consider their understandings about genetic testing to be weak, believing they "didn't know a lot" [P05] or that their "knowledge with regard to this whole area is really poor" [P13]. Even when their medical school curriculum included genetics, PCPs felt it didn't prepare them for WGS. One PCP stated, "Everything was monogenic disease risk. So the concept that there's something other than monogenic disease risk is something that I haven't quite fully digested yet" [P01]. The study education was generally considered helpful, although some PCPs felt it was more appropriate for physicians who already understood clinical genetics due to the terminology (e.g., "[PCPs] were not happy that there wasn't a primer… so that I could walk in that room and know what a GWAS is" [P10]) and nomenclature used (e.g., "They use a lot of abbreviations. They don't define them." [P05]).

While PCPs expressed apprehension about learning the concepts and language of WGS, they generally expected to be able to manage WGS findings effectively. Because PCPs felt

responsible for managing most aspects of their patients' wellbeing, all types of results were relevant, and responding to WGS was seen as little different from current obligations to respond to unfamiliar and novel information. PCPs wanted to be able to explain WGS results to patients and families, but recognized that they might not be the best physicians to act on them. "When it's like an oncologic thing and you don't know the detailed answer," summarized one physician, "You can say, 'Well, talk to your oncologist'" [P14].

Cardiologists, on the other hand, felt an obligation to respond competently to cardiac-related results. They were concerned about information they had not seen before, like polygenic risk predictions for cardiometabolic traits. Of frequent concern, however, was their responsibilities about findings unrelated to cardiology, which they considered incidental. Cardiologists, like PCPs, anticipated making numerous referrals, but felt uncomfortable determining when they were necessary. "You have all this other information," one cardiologist reported. "Whose responsibility is it to tell the patient?" [C07].

Motivations about Developing Proficiencies

By far, both cohorts cited developing proficiencies in WGS most often as a reason they enrolled in the study. They anticipated great value in disclosing actual WGS results to patients, believing "the more you do something, the better you become" [P14]. PCPs tended to discuss these benefits as learning about fundamentals of clinical genetics, such as the importance of family history and concepts such as penetrance and expression. Cardiologists also discussed the study as an opportunity to "relearn sort of the basics of genetics," [C07] but more often discussed aspirations of becoming proficient in applying WGS to patient care, such as "learning more about the entire way of approaching how we decide who to test and how to interpret the results" [C09]. Cardiologists were also more likely to address how WGS would introduce new challenges for a tool (i.e., genetic testing) they're using already, such as "how to deal with the informatics" [C02].

Integral to expectations about developing proficiencies was having support. Both cohorts anticipated "a lot more hand holding and instruction and support" [C18] than they would receive in non-research contexts. Many physicians wished that genetic counselors were incorporated more into the protocol to exemplify how clinical encounters should proceed. The GRC was appealing given common liability concerns about misinterpreting and miscommunicating WGS results. There was also recognition that feelings of unpreparedness and liability concerns would discourage physicians from engaging in genomic medicine, with one physician stating that PCPs, "Would just throw up their hands and say, 'You need to see a genetic counselor'" [P01].

Factors Affecting Preparedness

Concerns about preparation were often exacerbated not only by the complexity of WGS reports, but also by a lack of guidance on them. The majority of physicians felt the reports were suitably designed, but many wished they had concrete recommendations about clinical management. "I would love it if it said here, 'Get an echo, and then send to cardiologist'" [P05].

Nevertheless, providers believed they could respond appropriately if given time to prepare. Physicians expected to self-educate by reviewing scientific literature and genetics- and disease-specific websites. Physicians from both cohorts also wanted time to make use of support resources established for the study, stating "I will make it my business to make sure that I've met with the people at *the* Genetic [sic] Resource Center and discussed this or looked into it enough so that by the time I meet with the patient, I will feel comfortable" [C09]. Physicians from both cohorts also anticipated seeking advice from genetic specialists, particularly cardiologists who worked with genetic counselors in teams where they were "bouncing things off of each other" [C02]. Of note, physicians described concerns about policies that limit reimbursement for preparation time. "It takes a ton of time before and afterwards, and it's uncompensated time." [C03].

A lack of standards for the management of WGS results also left some physicians feeling uncomfortable. "We have guidelines [for other medical tests], and we know them or we look them up, and it's clear. It's easy. But this is not" [P05]. Providers anticipated seeking information about best practices from resources they were familiar with. For cardiologists, those tended to be genetics-specific resources like GeneTests and OMIM. For PCPs, these tended to be general medicine resources, like UpToDate® or journals like the New England Journal of Medicine. Providers anticipated seeking guidance on practical matters, such as billing for follow-up services, in addition to guidance about how to interpret results and information about specific conditions. Other practical issues of concern were the amount of clinical time it would take to discuss results with patients, how information would be stored and retrieved from medical records, and how clinical workflow might change.

DISCUSSION

This study summarizes the preparedness of PCPs and cardiologists to disclose WGS results to patients as part of a clinical research study. Findings reflected a mix of confidence and concern, with PCPs considering WGS another instance of an emerging technology that impacts their practice and cardiologists considering it a new version of a technology they were already using. Prior published data showed how physician participants expected WGS to be common in the future (27). We expand on those findings by highlighting how PCPs and cardiologists wanted to take advantage of WGS in the research setting to prepare themselves for this future, an educational strategy emphasized in a recent commentary (28). Both cohorts anticipated seeking guidance from study-created resources and from resources with which they were already familiar. The lack of guidance on WGS reports, time pressures, and a lack of standards contributed to concerns about disclosing and responding to WGS results.

Differences between PCPs and cardiologists were largely explained by their specialties' approaches towards medicine. Although PCPs considered their understandings of genetics to be limited, they had extensive experience receiving novel information as first line responders to patients' concerns. Cardiologists' familiarity with genetics, on the other hand, was offset by apprehensions about acting appropriately on new types of cardiac findings and making decisions about conditions unrelated to their specialty. Findings highlight how WGS training and infrastructure must address the needs of specific specialties. For PCPs, all

results were relevant and merited response. For cardiologists, non-cardiology results were incidental. Nearly all physicians reported benefitting from education, but some PCPs felt that our curriculum should have assumed less familiarity with the language and nomenclature of genetics.

Findings also identify institution- and policy-level factors that may facilitate or hinder genomic medicine. First, physicians may benefit from enhanced communication with laboratories. Desires for written guidance about how to respond to WGS reports may be inadvisable or even prohibited (29), given how findings should be considered alongside information that may be unavailable to laboratories (30). Nevertheless, discussions with laboratories can help physicians identify the phenotypic and personal or family history information that would support or refute the pathogenicity of a particular finding. These discussions may be particularly important for secondary or incidental findings, where false-positive rates are high (31) and physicians' knowledge may be especially limited.

Second, responding to WGS results may require more preparation time than other services, including targeted genomic tests. Reimbursement and malpractice policies may incentivize already-overburdened providers to reflexively refer patients to specialists, even when such referrals are unnecessary. If WGS becomes more routine in clinical care, such referrals may be particularly impractical given the limited number of genetic specialists (32).

Third, educational resources will need to be improved. Numerous initiatives have tried to empower providers to practice genomic medicine (19, 33–35), but resources that specifically address WGS are uncommon. Moreover, our findings show that physicians are likely to rely on resources with which they are already familiar. Popular resources such as UpToDate® have started to address WGS, but efforts will need to be expanded greatly (36).

Finally, healthcare professionals of all types will need to develop competencies in genomic sequencing. The high potential for incidental or secondary findings means that physicians who order WGS may be expected to disclose and act on results that are outside their area of expertise. It also highlights how physicians may be expected to respond to WGS information they had not ordered, but are receiving through a referral. Findings support prior calls to develop genomic competencies within clinical teams more broadly (37). Pharmacists will be expected to respond to pharmacogenomic findings, for example. Nurses may be particularly impacted because they typically collect the family history information used to interpret and contextualize WGS findings, and because they are at the forefront of health promotion efforts (38). Training programs and professional organizations have recognized the need for specialty- and profession-specific competencies, and groups are working to ensure that competencies are developed at a practical as well as conceptual level (39–42). The speed at which medical care is adopting WGS may necessitate engagement in these efforts soon.

One solution to many of these concerns may be to encourage patient care approaches that distribute the demands of WGS among teams rather than individual physicians (43). Inclusion of genetic specialists may be especially helpful. Genetic counselors are already part of team-based care in settings such as prenatal care, pediatrics, and oncology. Mirroring

this strategy in primary care may not only improve the care of individual patients, but also help in educating physicians and other healthcare professionals about genomic medicine.

A number of limitations merit discussion. The small sample size limited attempts to identify thematic differences by provider characteristics or survey measures. We enrolled a convenience sample of early WGS users from one academic hospital network, physicians who had time to participate, agreed to provide WGS to their patients and likely had positive attitudes about WGS. As one of the first clinical trials of WGS, the MedSeq Project provided enhanced physician support that may have mitigated concerns and may not be available in other settings. Further research will need to examine the beliefs and expectations of physicians who have less time or less favorable beliefs about WGS, and who practice at institutions where support is less developed.

Importantly, our data represents perspectives prior to using WGS. Physicians will be interviewed about their experiences at the end of the study, and future publications will address how well MedSeq Project participation fulfilled expectations about developing competencies and how the educational curriculum could be improved. We will also report how useful physicians found study-developed support tools, although early data suggests that they have contacted the GRC for assistance infrequently (i.e., about 5% of disclosures).

Nevertheless, our study identified important factors that affected how prepared physicians felt to use WGS, and barriers that are amenable to change. The potential for WGS to improve health outcomes is great, if physicians are prepared and empowered to use it.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Table 1

Responsibilities of the MedSeq Project's Genome Resource Center (GRC).

Task	Description
Advising	Physician participants called or emailed the GRC with questions about the reports or study procedures.
Safety Monitoring	GRC staff reviewed family history reports, sequencing reports, and disclosure session recordings to identify any information that physician participants miscommunicated to their patients and to identify important information that was ignored. Issues that introduced risks to patient participants' health were shared with physician participants immediately.
Education	During review of disclosure session recordings, GRC staff identified topics where physicians lacked confidence in their understandings. These topics were then addressed in educational sections of a periodic newsletter.

Table 2

Characteristics of enrolled physicians after education.

ID	Sex	Age	Race	Genetics Training	Frequency Discussing Genetics*	Knowledge Score (0–6)	Self Efficacy Score (1–5)	Self-rated Preparedness†
Primary Care Physicians								
P01	Male	64	White	No	Sometimes	5	3.3	Slightly
P05	Female	65	White	No	Sometimes	6	2.3	Slightly
P10	Female	53	White	No	Seldom	4	2.2	Not at all
P11	Male	64	Asian	Yes	Sometimes	5	4.0	Prepared
P13	Male	56	White	No	Seldom	5	4.5	Slightly
P14	Female	45	Black	No	Sometimes	5	3.5	Slightly
P15	Male	56	White	No	Seldom	3	3.0	Slightly
P16	Female	57	White	No	Sometimes	6	4.0	Slightly
P17	Female	41	White	No	Often	6	3.8	Slightly
P19	Male	39	White	No	Seldom	6	4.0	Slightly
P04§‖	Female	32	Asian	No	Seldom	4‡	3.8‡	Prepared‡
Cardiologists								
C02	Female	43	Asian	Yes	Almost always	5	3.3	Prepared‡
C03	Male	37	Asian	Yes	Often	6	4.7	Not at all‡
C06	Female	60	White	Yes	Almost always	5	3.5	Prepared
C07	Male	60	White	No	Seldom	6	2.0	Slightly
C08	Male	51	White	Yes	Often	6	3.7	Very
C09	Male	59	White	No	Sometimes	6	3.0	Slightly
C18	Male	48	White	No	Sometimes	5	2.7	Slightly
C21	Male	60	White	No	Sometimes	6‡	2.5‡	Prepared‡
C20§	Male	43	White	No	Often	4	3.2	Prepared

*Response options: never, seldom, sometimes, often, or almost always.

†Response options: very prepared, prepared, slightly prepared, or not at all prepared.

‡Reported from the pre-education questionnaire because post-education data was missing.

§Participant was not interviewed.

[||] Participant dropped out before completing the post-education questionnaire and interview.

Table 3

Primary and secondary themes related to physician preparedness for WGS use.

Theme	Description	Example
Primary		
Genetic literacy	Comments about understandings of genetics, genomics or WGS	"A lot of recessive stuff can have a phenotype, which I didn't know about." [P17]
Concerns about preparedness	Concerns about being unprepared to interpret, explain, or respond to WGS results	"I'm a little bit apprehensive about how much information may be provided and being able to convey the information in a clear manner." [P19]
Motivations about developing proficiencies	Statements about enrolling in the study to prepare for WGS use in the future	"[I] have followed the world of genomics and thought this would be a chance to learn more." [C21]
Secondary		
Previous experiences	Professional and personal experiences with genetic services	"I got emails from patients saying, 'Can you call my closest Quest Laboratory and order the BRCA gene for me?'" [P16]
Education sessions	Comments about the study-provided educational curriculum	"I actually learned a lot from those sessions, you know. Not only in terms of the reports themselves but how to look at mutations." [P15]
Response to reports	Comments about the WGS or family history reports	"[The reports have] about the right level of complexity." [C08]
Genome Resource Center	Comments about using the Genome Resource Center for assistance	"I'm going to use them as a way to educate me, initially, about what these conditions are." [C09]
Infrastructure	Comments about policies, programs, and tools that facilitate or hinder the use of WGS	"They won't then be able to store [WGS] reports in the medical record, because there is no format for doing that yet." [C08]
Genetic counselors' role	Anticipated use of genetic counselors to respond to WGS reports	"I would anticipate my leaning heavily on the genetic counselor." [C18]
Information seeking	Resources physicians expected to use to help interpret WGS reports	"… refer to the genetic counselor for a more detailed explanation for a specific condition." [P11]