

Primary Care Providers' Experiences With an Active Elective Genetic Testing Program

Health Education & Behavior
1–10
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DOI: 10.1177/10901981241266849
journals.sagepub.com/home/heb



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Abstract

Elective genetic testing (EGT) programs that provide pharmacogenomic information to guide medication management and screen for medically actionable disease predispositions are emerging in a number of health systems. Primary care providers (PCPs) are at the forefront of test initiation, patient education, and management of EGT results. However, little research has examined the experiences of PCPs in health systems offering clinical EGT. We conducted semi-structured interviews, a sub-study of the larger mixed-methods Imagenetics Initiative, with 16 PCPs at a health system in the Midwest with a clinical EGT program supported by provider education, automated clinical decision support, and enhanced access to genetic specialists. The purpose of these interviews was to understand perceptions about the benefits and barriers of implementing EGT in clinical practice. Thematic analysis indicated that EGT is conceptualized similar to traditional diagnostic services. PCPs were generally favorable toward EGT; however, targeted education did not dispel misconceptions about the goals, results, and limitations of EGT. Most PCPs endorsed the potential utility of EGT. Pharmacogenomic profiling was seen as having more immediate impact for patients than screening for monogenic disease risks. PCPs reported that they weighed discussions about EGT against time limitations and the need to prioritize patients' existing health concerns. Regardless of their education levels and familiarity with genetics, PCPs desired additional educational resources and greater access to genetic specialists. Our study provides unique insight into PCPs' experiences with clinical EGT in health systems that have adopted EGT and highlights the practical challenges and potential opportunities of EGT integration.

Keywords

elective genetic testing, population screening, personalized medicine, health care provider education, genetic education

Introduction

Genetic testing without a medical indication, elective genetic testing (EGT), is growing in use. This growth is driven by decreased cost, innovations in test delivery, and increased recognition that phenotype-driven risk assessment is insufficiently sensitive (East et al., 2019; Lu et al., 2019). EGT differs from diagnostic genetic testing (DGT) in that EGT is offered to apparently healthy patients rather than patients with a medical indication (Blout Zawatsky et al., 2023). Although most instances of EGT occur via direct-to-consumer services (Lu et al., 2019), concierge medical care (Goldberg, 2019), or research studies (Biesecker et al., 2009; Holm et al., 2018; Vassy et al., 2014; Zoltick et al., 2019), EGT is also available at an expanding number of health care systems (Anderson et al., 2021; Cochran et al., 2021; David et al., 2021; Lemke et al., 2020). Across all settings, the implementation of EGT will be carried out by health care professionals who did not formally

train in genetics and also have different educational/training backgrounds, including primary care providers (PCPs) and obstetrics/gynecology providers (OBGYNs). Little is known

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about how these health care professionals interact with EGT (Bean et al., 2021; Lemke et al., 2017).

Differences between EGT and DGT complicate its implementation. Genetic variants identified during EGT may be less penetrant than variants identified from phenotypically ascertained patients who receive DGT, and disease may present with a broader spectrum of mild or atypical phenotypes (Bean et al., 2021; Forrest et al., 2022; Hagenkord et al., 2020; Lu et al., 2019; Murray et al., 2020; Natarajan et al., 2016). There are limited data about the prevalence or penetrance of most rare genetic conditions in general populations, making it difficult to assess the sensitivity and specificity of EGT. In addition, counseling EGT patients with pathogenic variants about disease risk and management is nuanced, given that most diagnostic criteria and guidelines for genetic conditions assume the presence of a phenotype or family history suggestive of disease (Schwartz et al., 2021).

Real-world evidence about the experiences of providers in health systems with well-established clinical EGT programs is limited (Lemke et al., 2017; Paneque et al., 2016). To address this knowledge gap, we conducted semi-structured interviews with providers who effectively act as PCPs (a patient's primary point of medical contact) at Sanford Health (internal and family medicine, as well as OBGYNs). In 2014, Sanford Health enacted an "Imagenetics Initiative" (Internal medicine and genetics) that culminated in the launch of an EGT program available in primary care settings (Bell, 2017; Christensen et al., 2021; Petry et al., 2019). The goal of this study was to provide insight about the experiences of PCPs with EGT in a single health system that implemented EGT in tandem with education and clinical decision support for PCPs.

Method

This is a qualitative manuscript which is a sub-study to a much more broad, mixed-methods, research initiative; the Imagenetics Initiative.

Overview of Genetic Testing at Sanford Health

A multidisciplinary team that included a practicing PCP (C.H.) collaborated to design this study. Although we did not use a specific implementation framework to guide study design, analyses focus on contextual factors related to clinicians and health systems as proposed in the Genomic Medicine Integrative Research (GMIR) Framework. The GMIR Framework has been used to guide implementation studies of both rare genetic diseases and pharmacogenomic testing. It emphasizes how clinician attitudes interact with health care system factors (infrastructure, access to specialists, resources) and decision support to influence the adoption of genetic services.

Summaries of the Sanford Health System's infrastructure and education to support EGT in primary care have

Impact Statement

Our research details the experiences of, unmet needs, and potential misunderstanding of physicians, nurses, and physician assistants about the incorporation of elective genetic testing into primary care. Although clinicians at Sanford Health were required to engage in genetic education modules, had clinical decision support through the electronic health record, and had access to a large number of genetic specialists, they still advocated for a number of additional supports, continued education, and expressed misunderstandings of and barriers to the integration of elective genomic testing within their practice. We feel that findings from our study will help health system leaders and policymakers better understand the shortcomings of elective genetic testing programs and the needs of clinicians when incorporating this service into primary care.

been published previously (Blout Zawatsky et al., 2022; Christensen et al., 2021; Hajek et al., 2022; Petry et al., 2019). Briefly, efforts included growing Sanford's genetics workforce (including genetic counselors [GCs], medical geneticists) and expanding access to these specialists; identifying PCP "Physician Champions"; adapting electronic medical records system to store genetic test results; implementing clinical decision support for findings related to drug metabolism (pharmacogenomics; PGx) and genetic risk factors for diseases with well-established management guidelines (medically actionable predispositions, or "MAPs"); developing workflows for referring patients to genetic counseling and/or clinical genetics evaluations; and arranging genetics education for all health care and advanced practice providers at Sanford Health. From 2017 to 2019, providers completed quarterly educational modules with content and objectives summarized in Table 1. Providers hired after June 2019 completed an abbreviated two-module genetics education program rather than the formal eight-module genetics education.

In 2018, Sanford Health began offering the Sanford Chip, an EGT that provided PGx panel testing and optional genotyping for MAPs, to its adult primary care patients (Christensen et al., 2021). The EGT was advertised directly to patients and providers as an option to guide medication management and disease surveillance. Patients received messages through the electronic medical record, whereas regular clinical meetings addressed providers' questions about the use of EGT. Additional details about the provider education program, availability of genetic specialists, and genetic testing platform are provided in the Supplemental Materials.

Interview Participants and Study Design

Physicians, nurse practitioners, or physician assistants practicing in family medicine, internal medicine, or OBGYN were

Table 1. Content and Learning Objectives of Provider Education Modules.

Module and dates	Overview	Learning objectives
Module 1: What is genomic medicine?	Foundational material to better understand genomic medicine and how it can impact clinical practice	<ul style="list-style-type: none"> • Describe genomic medicine. • Interpret a pedigree. • Differentiate between genotype and phenotype. • Determine when to refer a patient for a genetic medicine consult. • Describe PGx and its benefits.
Module 2: Current applications of genomic medicine	Descriptions of genomic applications of precision medicine, including preemptive genomic screening	<ul style="list-style-type: none"> • Recognize genomic applications of precision medicine. • Classify the components of genomic medicine. • Describe the Sanford Chip and its clinical utility.
Module 3: The genetics of drug response	Clinical utility and application of PGx testing and the basics of drug metabolism	<ul style="list-style-type: none"> • Recognize the strengths and limitations of preemptive precision medicine. • Define PGx metabolizer types in the context of prodrug versus active drugs. • Identify the scientific organizations that create the guidelines for clinical application of PGx. • Recognize the components of the PGx test and utilize decision support tools. • Order PGx testing and apply results.
Module 4: Different types of genetic tests and specialists	Examples of genetic variation and types of tests used to identify each, along with types of genetic specialists available to offer support	<ul style="list-style-type: none"> • Differentiate between somatic and germline variation. • Summarize the different types of genetic testing. • Recognize the clinical application for each type of genetic testing. • Examine the clinical relevance of the genetic counseling process. • Distinguish the difference between genetic professionals. • Apply the principles of PGx to patient care. • Recognize cases in which PGx testing is appropriate. • Discuss the advantages and disadvantages of current approaches to PGx testing. • Recognize the components of PGx reports and utilize decision support tools. • Identify clinical resources related to PGx testing. • Summarize past efforts and current opportunities related to precision medicine. • Outline the spectrum of genetic changes, or variants, between Mendelian inheritance and common disease. • Characterize SNPs. • Appreciate the design and clinical utility of genome-wide association studies (GWAS). • Assess the clinical application of polygenic risk scores (PRS) to modify patients' clinical risk categories for more precise screening and treatment.
Module 5: PGx in patient care	PGx principles review and common case examples showcasing available clinical decision support tools (recorded video lecture)	<ul style="list-style-type: none"> • Describe the three main initiatives of Sanford Imagenetics with an emphasis on the Sanford Chip. • Delineate the Sanford Chip workflow for return of results. • Apply Sanford Chip results to clinical practice through case examples.
Module 6: The spectrum of genetic variants	Comparison of the genetics of Mendelian and common diseases with an introduction to the identification and analysis of single-nucleotide polymorphisms (SNPs) (recorded video lecture)	<ul style="list-style-type: none"> • Describe how a genetic diagnosis can aid patient care. • Evaluate cases in which referring a patient for a genetic medicine consult may be valuable. • Apply PGx testing results to medical management. • Discuss the value that the Coordination of Rare Diseases at Sanford (CoRDS) provides to patients, families, and researchers.
Module 7: Genetic screening and the Sanford Chip	High-level overview of Imagenetics with a focus on the return of results workflow for Sanford's precision prevention tool, the Sanford Chip	
Module 8: Using genomics to improve management	Case examples outlining how a genetic diagnosis improves patient outcomes and a brief description of Sanford's rare disease registry	

eligible for interviews, as these providers traditionally serve as PCPs at Sanford Health. Nurse practitioners in women's health and certified nurse midwives in any specialty were also eligible. Invitations to participate and two reminders were emailed to over 800 Sanford PCPs. Department meetings, educational luncheons, and the Sanford physician portal also advertised the study. PCPs who expressed interest received follow-up emails from the study team. Emails directed potential participants to complete a survey collecting information about their area and years of practice, gender, age, self-identified race and ethnicity, and contact information. The survey asked individuals about their familiarity with, enthusiasm for, perceived clinical utility of, and ease of access to the Sanford Chip. Eligible providers were then asked up to three times by email and/or phone to participate in semi-structured interviews. No incentives were offered for participation in the study.

Semi-Structured Interviews

Sixteen telephone interviews were conducted via telephone by GCs (A.M.H., D.M.P.) who were trained by a researcher with extensive qualitative research experience (L.J.). Interview guides were informed by review of relevant literature and focused on PCP characteristics, perceived benefits and barriers to EGT, and experiences with EGT (Supplemental Text 1). Interview questions were iteratively refined during training and approximately 10 pilot interviews with study team members who were also health care providers. Interview length ranged from 20 to 47 minutes.

Data Analyses

Interviews were recorded and transcribed prior to analysis. Grounded theory informed thematic analysis of interview transcripts in accordance with a six-step approach (Braun & Clarke, 2013, 2021). Thematic analysis included both inductive and deductive coding, with an initial codebook developed based on study goals that was iteratively updated until saturation of relevant themes was observed. All interview transcripts were double coded by two of three members of the research team (J.L.L., L.J., D.M.P.) using the finalized codebook, with discrepancies resolved through discussion. Analyses of coded transcripts were conducted using ATLAS.ti (ATLAS.ti Scientific Software Development GmbH, 2022). Interviewee demographics are presented in Table 2; this is a qualitative study of PCP experiences with EGT. Data regarding other aspects of the program and findings from the associated research program have previously been published (Bell, 2017; Blout Zawatsky et al., 2022; Christensen et al., 2021; Hajek et al., 2022; Massmann et al., 2023; Petry et al., 2019). The study protocol was deemed exempt from research participants by the Sanford Research Institutional Review Board.

Table 2. Characteristics of Interviewees.

Characteristic	N (%)
Specialty	
Family medicine	6 (37.5%)
Internal medicine	8 (50.0%)
Obstetrics and gynecology	2 (12.5%)
Role	
Physician	9 (56.2%)
Physician assistant	5 (31.2%)
Nurse practitioner	1 (6.2%)
Nurse midwife	1 (6.2%)
Years in practice	
<5 years	3 (18.8%)
5–9 years	6 (37.5%)
10–14 years	1 (6.2%)
15–19 years	2 (12.5%)
20+ years	4 (25.0%)
Gender	
Female	10 (62.5%)
Male	6 (37.5%)
Self-identified race and ethnicity	
White	13 (81.2%)
Asian	1 (6.2%)
Other	2 (12.4%)
Familiarity with Sanford Chip	
Not familiar	1 (6.2%)
Somewhat familiar	5 (31.2%)
Very familiar	10 (62.5%)
Enthusiasm about using the Sanford Chip in patient care	
Very or somewhat unenthusiastic	3 (18.7%)
Neutral	2 (12.5%)
Very or somewhat enthusiastic	11 (68.8%)
Perceived usefulness of Sanford Chip for patients	
Not at all or not very useful	2 (12.4%)
Somewhat useful	7 (43.8%)
Extremely or very useful	7 (43.8%)
Ease of Sanford Chip for the provider and their patients to access and use	
Very or somewhat difficult	4 (25.0%)
Neutral	5 (31.2%)
Very or somewhat easy	7 (43.8%)

Note. Characteristics of individual providers are summarized in Supplemental Table 1.

Results

Participant Characteristics

Of the 25 eligible screening instrument respondents, 16 (64%) participated in interviews, nine did not respond to scheduling inquiries. Participant characteristics are summarized in Table 2. All were aware of the Sanford Chip, although one participant was not familiar with the program. Ten of the interviewed clinicians reported being “very familiar” with the EGT program. Feelings about the program were mixed, with 11

Table 3. Additional Themes With Supporting Quotes.

Quote number	Interviewee	Theme	Quote
1	OBGYN-8, Nurse Midwife	Prompted referral to genetics	“If I see someone who’s had a family history of diabetes, cancers, I’ll say we have this Chip test that will tell us about your family history, and talk with a genetic counselor”
2	OBGYN-12, MD	EGT is at odds with traditional medical training	“The traditional medical teaching is you treat the patient, not the lab, you don’t just throw a bunch of labs at them and see what sticks. And so I think this kinda felt like it was an opposition to that whole principle.”
3	IM-23, MD	Thankful the screening is limited	“And so the more clearcut it is and there’s an action to it, I think helps,” reported one physician in internal medicine. “I have always appreciated that so far—the way that the Sanford Chip has been done.”
4	IM-16, PA	Inaccurate descriptions of uninformative results	“I would explain that to patients is there are some things that are popping up, but none of them are a clear indication to change our plan as we go forward.”
5	Fam Med-6, PA	EGT conversations can aid in shared decision-making	“there’s maybe a cluster of symptoms that concern me about a potential risk for the patients asking, not specifically for Chip but is there something else that I should be doing.”
6	IM-17, MD	PGx results can be helpful in medication adherence	“I feel like sometimes when they are struggling and they feel like none of the medications work, I can show them after the Chip test, like, okay, based on your genetics, this is what is safe. It is worth a try or consider it again . . . Some people feel like, ‘okay, that’s probably why my old one wasn’t working’.”

interviewees expressing some enthusiasm, two interviewees rating it as not or not very useful, and three reporting that it was difficult for PCPs and patients to access. Characteristics of individual interviewees are summarized in Supplemental Table 1.

Interview Findings

Four major themes emerged from interviews: (1) conceptualization of EGT as DGT, (2) a desire for more education, (3) considerations of utility, and (4) barriers to EGT.

Theme 1: PCPs Often Conceptualized Elective Genetic Testing as DGT

PCPs lacked familiarity with EGT for disease screening purposes. They often described the Sanford Chip as similar to diagnostic and screening modalities, such as imaging, which might reveal unanticipated findings or findings with limited clinical significance “kinda like giving a patient a CAT scan who’s asymptomatic” (OBGYN-12, MD). Many PCPs said they only spoke about the program when patients had personal or family medical histories suggestive of inherited disease, and in some of those cases, PCPs discussed how a referral to genetics may be appropriate (Table 3, No. 1). Some PCPs viewed discussions about the Sanford Chip as opportunities to re-address family histories of disease, including those who had declined targeted genetic testing, to “give them an opportunity to rethink it” (IM-17, MD).

PCPs’ tendencies to think about the Sanford Chip as DGT reflected a common sentiment that offering EGT to healthy patients is at odds with their training (Table 3, No. 2). This

discomfort was often linked with concerns that EGT could be a “fishing expedition” (OBGYN-12, MD), finding problems that did not need addressing. These concerns raised questions about possible financial obligations for downstream medical evaluations. “That’s no longer preventive care and now it’s not covered by insurance” (OBGYN-12, MD). Other PCPs applauded the program for limiting its scope to PGx information and screening for a small set of conditions with clear management guidelines (Table 3, No. 3).

Theme 2: Misconceptions About EGT and Desire for Additional Education

Despite mandatory genetics education and creation of educational resources, uncertainties about EGT were common. These included a lack of understanding about the limitations of the Sanford Chip (e.g., genotyping array not being as accurate or comprehensive as next-generation sequencing, “uninformative” results not ruling out all genetic risks). One PCP noted “I probably would need to educate myself more to actually even be able to educate patients on the limitations to it” (Fam Med-6, PA). In most cases, PCPs were not confident about topics such as how the test may evolve to address additional drug–gene interactions, how to identify the most appropriate populations for testing, and the implications of uninformative MAP results. Almost every interviewee endorsed the need for additional, ongoing education, including example use cases for testing and language for disclosing results. In fact, some PCPs discussed concerns that they may not have discussed those aspects with patients well previously, stating that they “didn’t know there were any risks to it” (OBGYN-8, Nurse Midwife).

In addition, a number of PCPs discussed the need for ongoing updates about the program. Some expressed concerns that they were not always aware of what conditions it tested for. A common suggestion was the continued use and adaptation of “cheat sheets,” frequently asked questions and printed materials they could refer to as needed. One PCP, for example, desired a reference sheet about PGx and relevant medications that she could “just put it in my office and quickly look through if there is an updated version” (IM-17, MD). The need to practice evidence-based medicine was emphasized by most PCPs, and several encouraged the interviewer to develop systems to update clinics about how EGT improved patients’ health. In the absence of such information, some PCPs stated that they may forego discussing the program with patients. Several PCPs felt uncomfortable about being an “advocate” for the Sanford Chip, stating “I have no idea what has Sanford Chip done for the patient’s health (Sic)” (Fam Med-18, MD).

Several PCPs raised concerns about possible insurance implications for patients when genetic predispositions were identified. Some PCPs were aware of protections such as the Genetic Information Nondiscrimination Act, but there was confusion about what this legislation did and did not cover, as well as concerns about whether insurance would cover follow-up procedures in asymptomatic patients. This confusion led to uncertainty about how to manage MAP results, with one PCP stating, “I’ve been told not to put certain things on charts because the insurance won’t cover something” (IM-22, MD).

Another common topic of uncertainty about EGT was how to explain uninformative MAP results. PCPs mentioned that a common patient question about MAP results is, “Why is it not positive or negative?” (IM-28, MD). Some PCPs were unaware that this language was deliberately used because the Sanford Chip’s use of array-based testing does not identify all pathogenic variants, which led to inaccurate descriptions of results (Table 3, No. 4).

Theme 3: Perceptions of Utility

Nearly all PCPs interviewed believed that the Sanford Chip provided at least some utility. PCPs often viewed conversations with patients about the program as opportunities to build relationships and provide more holistic care. Some PCPs addressed how conversations about the program could aid in shared decision-making about specific health decisions (Table 3, No. 5). Several PCPs spoke about the difficulty of convincing patients to complete recommended health screenings (e.g., colonoscopies) or to adopt healthier lifestyles. These respondents often saw the Sanford Chip as a tool to revive these conversations, particularly if patients were found to have a MAP “I think that might be something if we could convince people to do some genetic testing, you know, and tell them, look, hey you have this that would predispose you to cancer” (Fam Med-6, PA). PCPs often felt discussion of EGT allowed PCPs to emphasize the importance of traditional screening services, providing “another discussion point to try to get more people

to do it” (IM-16, PA). Other PCPs expressed limited enthusiasm for discussing genetic testing with patients who were already following evidence-based guidelines for screening. PCPs also addressed how discussions about the Sanford Chip prompted PCPs to collect more comprehensive personal and family medical histories of their patients.

Nearly all interviewed PCPs expressed that PGx results were more likely to be immediately helpful to patients than information about MAPs. Many PCPs expressed they had only ever received actionable PGx results about their patients, not MAPs. Some stated that they ordered the Sanford Chip for their patients to aid in prescription management or to explain why past medications had been ineffective. PCPs often discussed how Sanford Chip results may be helpful for persuading patients to adhere to or change medications (Table 3, No. 6).

Theme 4: Barriers to Offering EGT

The most prevalent barrier to discussing EGT with patients was a lack of time. One PCP summed up the lack of time by expressing if interest in the Sanford Chip is “one of seven complaints at their annual exam, it’s gonna be hard to get to everything . . . so that’s really patient dependent” (OBGYN-12, MD). Several PCPs mentioned that the extended nature of conversations about the Sanford Chip could compound the demands of patient care. PCPs also discussed how clinical decision support alerts triggered by PGx results competed against other clinical decision support alerts and how addressing Sanford Chip findings needed to be prioritized against patients’ questions and more immediate health needs. EGT was typically considered a low priority if patients had acute health needs, were not regularly engaging in conventional screening, or were living unhealthy lifestyles. Addressing Sanford Chip findings could be particularly challenging when PCPs perceived an overall lack of guidance about how genetic findings should affect patient management.

These feelings were compounded by how PCPs often felt an overall lack of motivation among patients to initiate health changes even following a positive genetic test result. “I think that patients are pretty hard to motivate . . . I think the ones who want to make changes are the ones who are living pretty healthy to begin with” (IM-22, MD).

Discussion

This study provides insight into the perceptions and experiences of PCPs who practiced in a health system with a large clinical EGT program. Specifically, our study revealed four main themes:

1. Although many were enthusiastic about the program, PCPs often did not recognize EGT as distinct from DGT and felt that EGT in healthy people was at odds with our current approach to health care.

2. Misunderstandings and uncertainties about the intended uses of EGT and how to counsel patients about results illustrate how front-end provider education and support need to be complemented with continued genetics education.
3. EGT was perceived as generally useful, even when it was only a topic of discussion with patients. PCPs generally recognized the utility of PGx testing, but the utility of screening for MAPs was less obvious.
4. Implementation of EGT in primary care settings faces obstacles, including the desire to prioritize issues that PCPs may consider more important, such as existing medical needs or disease prevention; time limits to medical visits; and concerns about insurance and financial implications for patients.

The overall goals of offering the Sanford Chip were twofold: (1) make PGx information more available to improve medication management and (2) identify MAPs in patients who otherwise would never know they had significant risks for preventable conditions. Our four main findings have policy and practice implications relevant to both of these goals.

The nontargeted provision of the Sanford Chip and the long history of genetic testing as a diagnostic service rather than a preventive medicine service make it unsurprising that PCPs in our sample conflated the two approaches. The finding that most PCPs do not recognize EGT as different from DGT demonstrates that additional education about EGT specifically may be necessary to enable PCPs to confidently discuss this kind of testing with patients. Education should highlight that most individuals who have MAP variants are unaware of their genetic risk status and do not meet criteria for targeted genetic testing (Blout Zawatsky et al., 2021; Grzymiski et al., 2020; Manickam et al., 2018), based on personal or family history. Such education would need to encourage a shift in mind-set from treating illness to disease prevention to make clearer the benefits and limitations of preemptive EGT. Despite improvements in perceived preparedness following initial education (Hajek et al., 2022), PCPs at Sanford Health and other health systems may benefit from ongoing reinforcement of certain topics that are delivered in concise, efficient ways. As the evidence base grows, continuing provider education should include updates about practice guidelines for EGT and data about its impact on patient outcomes.

In our study, PCPs discussed EGT for PGx as intuitively useful, while they viewed EGT for MAPs as less obviously useful. It is likely that the PCPs' perceptions about the relative utility of PGx results compared with MAP results reflects, at least in part, how the program was described and the frequency of actionable findings. Well over 90% of patients who completed the Sanford Chip program have atypical PGx results compared with about 4% of patients for MAP results (Christensen et al., 2021). Although the likelihood that PGx testing provides a high-impact benefit is extremely low, given that drug-gene interactions with life-threatening implications

depend both on patients having a rare genotype and the relevant medication being ordered. PCPs also expressed concerns that using language describing a lack of MAP results as “uninformative” rather than “negative” was confusing for patients. The different attitudes of PCPs toward PGx and MAP testing raise questions about the implications of bundling these two types of information into one test. Some PCPs who are favorable to PGx testing may be reluctant to endorse the Sanford Chip due to concerns about counseling patients about MAP results. To maximize PCPs' willingness to engage with EGT, health systems may want to consider offering PGx testing and MAP screening as independent elective services when offered in primary care settings.

The most common reasons PCPs in our sample did not offer the Sanford Chip to patients were competing priorities and limited time. Although these have been consistent barriers to the introduction of new primary care programs (Korownyk et al., 2017), it is important to note that Sanford's efforts to address these concerns may have been insufficient. Despite coordinated web-based provider and patient education and robust clinical decision support (Blout Zawatsky et al., 2022), most PCPs highlighted these barriers to the Sanford Chip. EGT programs may benefit from additional patient educational resources being available before certain visit types instead of being offered only during informed consent to further alleviate time demands. Primary care settings may need additional supports to educate patients about EGT for PGx and MAP screening, such as longer visits, better online patient education, dedicated EGT educational staff, and report formats that can better enhance patients' understanding of their results (Farmer et al., 2020; Haga et al., 2014).

The insight that PCPs may have too many competing priorities to discuss EGT consistently does not mean that EGT should remain in the sole purview of genetics professionals and direct-to-consumer companies. At Sanford Health, patients could request testing independently, and evidence suggests that most of the information patients need about EGT prior to testing can be conveyed effectively using written materials or interactive electronic media (Blout Zawatsky et al., 2023; Green et al., 2001). Admittedly, patients will continue to turn to PCPs for help interpreting and acting on test results. As such, continuing provider education should focus on honing the skills and knowledge PCPs need to interpret, discuss, and develop a patient care plan based on EGT results. This will allow genetics specialists to reserve their skills for consultations about difficult cases (Maxwell et al., 2020). Our results suggest that honing the division of labor between digital educational tools, PCPs, and genetics specialists will be important to pave the way toward precision health care for all.

PCPs in our sample were also concerned about the potential of EGT to lead to genetic discrimination. To date, evidence that patients have experienced insurance or employment discrimination based on genetic results has been anecdotal, even in high-risk populations (Golinghorst & Prince, 2020; Wertz, 2002). Nevertheless, the lack of federal protections

on the use of genetic test results by insurers to affect life, disability, and long-term care insurance remains among the most-cited concerns of patients and PCPs alike (Hauser et al., 2018; Robinson et al., 2016). Given the aforementioned time constraints, PCPs may not have adequate time to have discussions about weighing the potential medical impact of EGT against potential discrimination based on EGT results and the relevant legal protections. Digital, patient-facing educational tools may be useful in addressing this need.

Limitations of this study include a small sample size of individuals who tended to have positive attitudes about EGT. Social desirability bias may have occurred during interviews and led participants to provide more positive comments about EGT than they actually felt. PCPs may not have received the same genetics education, depending on their hire date as the original eight-module education was reduced to two. In-person access to genetic specialists varied by location and time, especially during parts of the COVID-19 pandemic. Data came from a single Midwestern health system with limited ethnic diversity in its patient population.

Conclusion

For EGT programs to succeed, PCP educational efforts need to differentiate DGT versus EGT and highlight how to use EGT findings appropriately. Our research highlights challenges to EGT implementation that warrant consideration from other EGT programs. Current and future EGT programs should help PCPs balance discussions about EGT with the myriad other demands they encounter and address potential discrimination concerns. These challenges may be particularly important to address as additional mechanisms for obtaining EGT expand, including return of results from large-scale initiatives such as the All of Us Research Program (The “All of Us” Research Program Investigators, 2019).

Acknowledgments

The authors acknowledge the efforts of providers in the Sanford Health system who contributed to the work of the Imagenetics Initiative.

Declaration of Conflicting Interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The author(s) disclosed receipt of the following financial support for the research, authorship, and/or publication of this article: A monetary gift from Denny Sanford allowed Sanford Health to grow their workforce and begin the Imagenetics Initiative and also subsidized the cost for EGT (\$49). R.C.G., K.D.C., E.S.Z., M.R.H., J.L.L., and C.B.Z. were supported by a research grant from Sanford Health. K.D.C. was also supported by NIH grant K01-HG009173. C.B.Z. is compensated from Atria to coordinate and provide genomics education. R.C.G. has received compensation for advising the following

companies: AIA, Allelica, Atria, Fabric, Genome Web, Genomic Life, Verily, and VinBigData and is cofounder of Genome Medical and Nurture Genomics. L.J. is supported by The Intramural Research Program of the National Cancer Institute. D.M.P. and A.M.H. are paid employees of Sanford Health.

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Supplemental Material

Supplemental material for this article is available online.

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