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Primary Care Physicians' Experience and Confidence with Genetic Testing and Perceived Barriers to Genomic Medicine

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Abstract

Purpose: Genetic testing is progressing towards use of patients' genomes for personalized medicine. Primary care physicians (PCPs) may use genetic tests to screen and assess risk. However, PCPs' current preparedness for the expanding integration of genetics into practice is uncharacterized. We examined primary care physicians' perceptions of and experience with genetic testing.

Methods: An anonymous survey was mailed to PCPs across three regional health networks querying opinions of, experience with, confidence in, and perceived barriers to genetic testing.

Results: The survey response rate was 37.8%. Respondents believed learning about new genetic advances was important to clinical practice (67.0%). A minority (19.0%) had ordered genetic testing in six months, with cancer risk testing the most frequently ordered. Respondents were not confident in the skills required for using genetic testing in practice. Few respondents felt that they had time to counsel about genetic risk (9.5%) or that most patients could comprehend the concept of risk (27.0%).

Conclusions: Primary care physicians had a high opinion of using genetic testing in medicine, but reported little experience or confidence incorporating genetic testing into practice. A majority perceived time constraints and patient comprehension as barriers. These data demonstrate a need for genetics educational resources for physicians and patients.

Keywords: Genetics; Primary care; Family medicine; Personalized medicine; Genomics

Abbreviations

PCP: Primary Care Physician; DTC: Direct to Consumer

Introduction

From the completion of the Human Genome Project in 2003 to the recently mandated withdrawal of the genome screening products of 23 and Me™ (Mountain View, California) there has been exponential development in health-related genomic information [1]. A glimpse of the potential benefit of genomic information in healthcare has been realized in targeted chemotherapy [2], tailored warfarin therapy [3], diagnosis of Mendelian disease [4], and expanded newborn screening [5]. These successes have heralded the development of "personalized medicine," which incorporates a patient's unique genomic make-up into individualization of disease prevention and treatment

Genomic science is developing rapidly, but much of basic science research remains too ambiguous to translate to clinical medicine. At the benchtop, DNA sequencing is still too inaccurate to use on a large scale, and once a DNA sequence is completed, the significance of each discovered variant is still largely unknown [6]. Despite these barriers, the public's interest in genetics fueled the development of companies like 23 and Me™, a company that provided Direct-to-Consumer (DTC) genetic tests. In November, 2014, the FDA suspended the company's health-related genetic reporting services due to lack of validation of the genetic tests and their uses [7], but the possibility of new companies arising remains.

Primary Care Physicians (PCPs) play a unique role in medical genetics in that they will likely be the first providers to assess patient need for genetic testing [8]. PCPs are also the most likely physicians to be presented with DTC test results given their long-term relationships with patients [9-10]. PCPs have been anticipating the clinical impact of genetic tests in their practice since before the completion of the Human Genome Project [11]. Since then, many PCPs overall continue to note that they lack confidence, knowledge, and experience with genetic tests despite their anticipation [12-14]. These PCPs identified a need for increased education for themselves to reliably interpret test results [15-20], access to genetic counselors for referral [21], and increased education for the public to prevent anxiety and misinterpretations of results [22-23]. Further, they identified that ethical controversy exists over incidental findings, informed consent, and the ever-changing nature of genetic knowledge [24-27].

In 15 years of study into this area, primary care providers have reported that they perceive themselves deficient in assimilating the skills that they feel are necessary to participate in genomics medicine. The purpose of our study was to determine whether PCPs have

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improved in experience, confidence, and ability to order genetic testing since the recent surge of clinically relevant tests and well-publicized rise and fall of affordable DTC genetic tests.

Methods

Survey population and recruitment

Three regional networks of PCPs in the Delaware Valley participated in the study: 1) Jeff Care, an organization of PCPs affiliated with Thomas Jefferson University; 2) the Lehigh Valley Health Network, including its Practice-Based Research Network; and 3) the Christiana Care Health System and the Delaware Academy of Family Physicians.

Physicians were invited to participate in January, 2013. A postcard with the investigators' signatures and respective health system logos was mailed to every provider within the three networks to request his or her participation. Two weeks later, a packet was mailed containing a cover letter describing the project in detail, a link to an online version of the survey, a printed copy of the survey, a stamped and addressed return envelope, and a $5 gift card as remuneration for their time. The providers were informed that their surveys would remain anonymous. Consequently, non-responders were not identifiable and no follow-up emails or letters were sent.

Survey design

Investigators created a novel survey through an iterative process consisting of 20 questions, 5 of which had multiple sub-questions for a total of 50 answerable units. Thirty-four questions used a Likert scale, five were dichotomous (yes/no), six had multiple choices, three were "check-all-that-apply," and two were free text. All "check-all-that-apply" questions also included an "other" option with an accompanying free text box for description. The survey allowed participants to skip questions.

The survey consisted of five informal sections: 1) Demographic and practice information; 2) Opinions regarding genetic testing; 3) Experience with genetic testing; 4) Confidence in tasks in evaluating and managing genetic disease; and 5) Perceived barriers to implementing genomic medicine.

Funding and approval

The study was funded through an internal grant through the Dean's Office of Sidney Kimmel Medical College. The study was reviewed and approved by the Institutional Review Boards representing each of the participating health networks.

Data analysis

Survey data were entered into Excel for analysis. Personal characteristics were described as frequencies and percentages. The personal characteristics included age, gender (male, female), practice type (academic/private, rural/suburban/urban), specialty (family medicine, internal medicine, other), affiliation (Jeff Care, LVHN, CCHS/DAFP), and Electronic Medical Record (EMR) use.

Data were evaluated by descriptive statistics and comparisons of categorical responses from different groups were made using 2x2 contingency tables and Fisher's two-tailed exact test. Descriptive statistics include non-responder rates.

Table 1: Demographics of the Respondents.

	n	%
Gender		
Male	166	54.1
Female	141	45.9
Age		
25-34	48	15.6
35-44	78	25.3
45-54	86	27.9
55-64	79	25.6
65-74	11	3.6
≥ 75	6	1.9
Clinical Role		
Family Medicine Physician	204	65.8
Internal Medicine Physician	70	22.6
Other	36	11.6
Practice Setting		
Rural	20	10.0
Suburban	108	54.0
Urban	66	35.0
Multiple	22	11.0
Practice Type		
Academic	92	29.2
Private	106	33.7
Both	12	3.8
No option chosen	105	33.3
Healthcare Affiliate		
JeffCare	68	22.3
CCHS/DAFP	129	42.3
LVHN	108	35.4
Use EMR	275	89.0

Results

Survey response

A total of 833 postcards/surveys were sent to PCPs within the three networks. Of these, 315 surveys were returned resulting in an overall response rate of 37.8%. Less than a quarter (23.2%) of responders wrote free text box answers, which were mostly used to further describe their practices and did not contribute to the results.

Demographics

Slightly more than half of the respondents were male (54.1%) or between the ages of 45 and 64 (53.5%). Regarding specialty, 65.8% were family medicine physicians, 22.6% were internal medicine physicians, with 11.6% reporting other specialties including pediatrics, emergency medicine, geriatrics, and palliative care. Additional respondent demographic characteristics are listed in Table 1.

Opinions of genetic testing

Participants were asked their opinions regarding genetic testing.

Table 2: Primary care physicians' confidence in ability to perform evaluation and management tasks for diseases with a genetic basis.

Confidence	None	Low	Some	High	Very High
Take a thorough family history	10.2%	21.3%	46.0%	17.8%	3.5%
Counsel patients about risk reduction strategies based on the results of genetic testing	12.4%	27.0%	32.1%	19.4%	7.6%
Assess risk of hereditary disease	11.1%	30.2%	42.2%	11.7%	2.5%
Build a family medical history pedigree chart	13.7%	28.9%	33.7%	17.8%	4.1%
Counsel patients about whether or not to have genetic counseling	10.5%	36.2%	39.4%	11.1%	1.3%
Discuss risks, benefits, limits to genetic testing for hereditary cancers	19.4%	38.1%	32.4%	7.9%	0.6%
Order genetic testing for hereditary cancers	23.2%	45.4%	23.8%	4.8%	1.3%

Table 3: Confidence of PCPs based on experience. Percentage of respondents who had "some," "high" or "very high" confidence in ability to perform tasks for genetic-based disease if they had or had not ordered genetic testing or consulted a genetic counselor in the past 6 months.

	Ordered Genetic Testing in 6 mo		Consulted with genetic counselor in 6 mo	
	Yes	p-value	Yes	p-value
Take a thorough family history		**0.0469**		**0.1251**
Confident (n=211)	22.3%		20.8%	
Not Confident (n=103)	12.6%		13.6%	
Build a family medical history pedigree chart		**0.1127**		**0.0125**
Confident (n=174)	22.4%		23.6%	
Not Confident (n=140)	15.0%		12.1%	
Assess risk of hereditary disease		**0.0013**		**0.5587**
Confident (n=177)	25.4%		19.8%	
Not Confident (n=137)	10.9%		16.8%	
Order genetic testing for hereditary cancers		**0.0001**		**0.0167**
Confident (n=93)	35.5%		26.9%	
Not Confident (n=221)	12.2%		14.9%	
Discuss risks, benefits, limits to genetic testing for hereditary cancers		**0.0082**		**0.0379**
Confident (n=128)	26.6%		24.2%	
Not Confident (n=186)	14.0%		14.5%	
Counsel patients about whether or not to have genetic counseling		**0.0016**		**0.0005**
Confident (n=162)	25.9%		25.9%	
Not Confident (n=152)	11.8%		10.5%	
Counsel patients about risk reduction strategies based on the results of genetic testing		**0.0054**		**0.0047**
Confident (n=185)	24.3%		23.8%	
Not Confident (n=129)	11.6%		10.9%	

Approximately two-thirds of respondents agreed that learning about new advances in genetic testing was important to their practice (67.0%). Slightly less agreed that the genetic risk contributes a clinically meaningful portion of overall disease risk (64.8%). Very few agreed to the statement that physicians should consider genetic testing with all patients (15.9%).

Experience with genetic testing

A minority of respondents had ordered genetic testing (19.0%); the most frequent tests ordered were cancer risk testing (n=29), preconception/prenatal testing (n=11), and pharmacogenomic testing (n=9). A minority of respondents had consulted with a genetics counselor (18.4%) in the past six months, with the most frequent consultations for cancer risk testing (n=49) and prenatal testing (n=11).

The majority of respondents had initiated a discussion about genetic testing less than monthly (80.6%). Similarly, 88.6% of respondent's reported being asked about genetic testing by patients less than monthly.

Confidence in genetics and genetic testing

Participants were asked how confident they were in their ability to perform seven tasks in evaluating and managing diseases with a genetic basis (Table 2). Physicians were most confident in "ability to take a thorough family history" (67.3%) and least confident in "ability to order genetic testing for hereditary cancers" (23.8%).

Physicians aged 25-44 were more confident than physicians aged 45-54 in their ability to take a thorough family history (73.8% vs. 60.5%, $p<0.05$), build a family medical history pedigree chart (62.7%

Table 4: Percentage of respondents who had "some," "high" or "very high" confidence in counseling about a specific genetic disease or screening panel.

	Some	High	Very High	Total
Newborn Genetic screening	24.7%	57.1%	11.4%	93.2%
Hereditary non-polyposis colorectal cancer	38.2%	22.0%	0.6%	60.8%
MEN syndromes	36.6%	16.5%	3.6%	56.6%
Long QT syndrome	32.6%	16.8%	3.2%	52.6%
Hemochromatosis	30.0%	14.5%	3.9%	48.4%
Pharmacogenomics	37.1%	9.4%	1.3%	47.7%
Hereditary breast and ovarian cancer syndrome	34.9%	10.4%	0.7%	45.9%
Factor V Leiden	27.9%	9.4%	1.6%	39.0%
Prenatal genetic screening	24.1%	6.2%	1.0%	31.3%
Huntington Disease	23.6%	6.5%	0.3%	30.4%
Familial adenomatous polyposis	21.4%	4.9%	0.3%	26.6%

Table 5: Primary care physicians' evaluation of barriers to incorporating genetic risk assessment in primary care. Percentage of physicians agreeing with the following stand-alone statements.

	Agree	Strongly Agree	Total
I have enough time to counsel patients about genetic risk	9.2%	0.3%	9.5%
Most patients can understand the concept of genetic risk	25.7%	1.3%	27.0%
In my practice, I have adequate resources to help patients understand genetic risk	4.8%	0.0%	4.8%
My hospital or network has adequate resources to help patients understand genetic risk	24.4%	5.4%	29.8%

Table 6: Percentage of primary care physicians who find the following barriers to discussing the results of DTC genetic test as "somewhat" or "very" important.

	Somewhat Important	Very Important	Total
Inadequate Knowledge (self)	29.2%	54.5%	83.7%
Lack of Time	32.5%	41.7%	74.2%
Potential ethical dilemmas	33.7%	35.6%	69.3%
Lack of current relevance to medical decisions	34.1%	32.5%	66.6%
Lack of Reimbursement	25.2%	24.6%	49.8%

vs. 48.8%, $p<0.05$), and discuss risks, benefits, and limits to genetic testing for hereditary cancers (46.8% vs. 32.6%, $p<0.05$).

Physicians who had higher confidence ordered more genetic testing and were more likely to consult with a genetic counselor (Table 3). Physicians who had higher confidence more often agreed that genetic testing is important to their practice (all tasks, $p<0.0001$) and that genetics contributes a meaningful portion to disease (all tasks, $p<0.05$, except "order genetic testing for hereditary cancers" p=0.289).

Confidence did not vary significantly across practice type (academic/private) or setting (rural/suburban/urban).

Confidence in counseling on common genetic diseases in the primary care setting

Physicians were surveyed on how confident they were in counseling patients in the outpatient setting about genetic issues relevant to primary care (Table 4). The vast majority of physicians were confident in counseling about newborn screening (93.2%). Physicians were least confident in counseling about Familial Adenomatous Polyposis (26.6%).

Barriers to incorporating genetic/genomic testing into practice

Respondents were asked about barriers to incorporating genetic/genomic testing into their practice in two different ways.

First, they were asked how much they agreed with statements about potential barriers to the routine incorporation of genetic testing into their clinical practice (Table 5). Very few respondents believed they had enough time to counsel patients about genetic risk (9.5%) or that patients could understand the concept of risk (27.0%). Very few physicians felt that they had adequate resources in their practices to help patients understand genetic risk (4.8%); somewhat more respondents felt that their hospital or network had such resources (29.8%).

The second item was a clinical scenario with a patient wishing to discuss Direct-to-Consumer (DTC) genetic test results. Respondents rated how important various barriers were to discussing the test results (Table 6). The most important barrier was a personal sense of inadequate knowledge (83.7%). The potential barrier that was identified as least important was lack of reimbursement (49.8%).

Discussion

A majority of primary care physicians in our study had a favorable opinion of genetic testing, but little collective experience ordering genetic tests, consulting genetic counselors, or initiating

discussions of genetic testing with patients. Other studies of primary care physicians report a similarly positive opinion of genetic testing. However, the rate of ordering genetic testing or consulting with a genetic counselor in our study is less than half the rate in other studies [15, 28-29], a finding that may be attributable to the specialty mix of our population. Although our study found no significant difference between the specialties in percentage who had ordered genetic testing, previous research has demonstrated that family medicine physicians order fewer genetic tests than internal medicine physicians, opting instead to refer for testing [29-30].

The most frequently cited reason for ordering both genetic testing and genetics consultation was for the assessment of cancer risk. This is consistent with prior surveys indicating that breast and ovarian cancer risk assessment are the most common reasons PCPs order genetic testing [15, 28]. Most PCPs have familiarity with the increased risk of breast and ovarian cancer in a patient with first-degree relatives affected and has developed confidence in recommending genetic testing for appropriate patients [12-14]. Although primary care physicians are also aware of the genetic basis of many other hereditary disorders, they demonstrate low levels of knowledge and confidence in recognizing clinical scenarios where the testing is indicated [31-34]. Electronic medical systems may offer help with the integration of this new technology into practice. Our respondents reported a high rate of EMR use, reflecting national trends. Electronic medical records could potentially be programmed to incorporate genomic information and detailed family pedigrees into decision support systems for ordering genetic testing [35]. However, genetic and family pedigree information would have to be collected in a standardized fashion before such a system could be debuted successfully. Without such a system, if PCPs feel uncomfortable with cancer genetics, it may be best to refer to genetic counselors for optimal evaluation for the ordering and interpreting of genetic tests.

Our finding that physicians who felt confident in these tasks were more likely to have ordered genetic testing or consulted with a genetic counselor in the past six months is consistent with previous studies [15,17,28]. Physician confidence in these tasks also correlated with the beliefs that genetic testing is important and that genetic risk contributes to disease risk, reinforcing previous findings [18].

Younger physicians in our study (aged 25-44 years) reported more confidence than physicians aged 45-54 on a few genetic testing tasks. In previous studies, the effect of age or year of graduation on confidence levels has varied from no difference [15,19] to increased confidence in physicians 50 years and older [17,18]. While younger physicians score higher when tested for knowledge about genetics [36-37], they perceive that their ability to apply that knowledge is low, resulting in lower confidence, at least at the beginning of the learning curve. Despite the issue of low confidence, PCPs have previously adapted successfully to integration of new technologies. It seems likely that with increased education and consultation with genetics experts, PCPs will learn how to incorporate genetic testing and interpretation into their practices.

Physicians in our study reported having the highest confidence in counseling about newborn screening among all the genetic testing options. This is in contrast to recent research in which only 16.5% of Canadian family medicine physicians and 62.6% of Canadian pediatricians report feeling confident in their ability to explain newborn screening results to parents [38]. These disparities may reflect differences in physician training and clinical experience in Canada versus the United States.

Most PCPs identified lack of time and patient comprehension as impediments to counseling about genetic risk. When presented with a DTC testing scenario, PCPs added inadequate knowledge (self), lack of relevance, and potential ethical dilemmas to the list of barriers. Lack of time is the most commonly cited barrier to discussing genetic tests and results by PCPs; lack of knowledge, lack of clinical relevance, and moral/ethical concerns are also frequently mentioned [17-18, 39]. One recent survey of the public found that patients generally have good recall of genetic testing results and do not have a deterministic view of genetics. However, these findings were disproportionately more prevalent in patients with higher levels of education and higher socioeconomic status [40] and may not be generalizable. Other studies have concluded that there is public misinterpretation of the role of genes in disease, how genetic disease is transmitted, and what a positive genetic test means [9, 41-42].

The physicians in our study did not feel that their practices had resources to improve patient comprehension, and only three out of ten agreed that their hospitals or networks had such resources. Genetics counselors could potentially fill this niche, but there is great variability in PCPs' ability to make referrals to professionals with this skill set. Much of this variability can be attributed to practice setting. A recent study found that while 82.3% of PCPs based in academic medical centers reported that had access to genetics specialists for their patients, only 47.1% of community-based PCPs did [43].

Less than half of PCPs thought that lack of reimbursement was a barrier to discussion about DTC genetic test results. A previous study also found that PCPs were concerned about the costs associated with DTC genetic tests, but their concern was for the financial burden on their patients, not their own reimbursement [17]. Primary care physicians frequently provide counseling and management services that are not reimbursed [44], but they may be underestimating the amount of time it could take to explain results associated with indeterminate future risk.

Limitations of this study include potential response bias, since we were unable to determine how respondents might have differed from non-respondents. Participation may have been influenced by interest or knowledge about genetics. Additionally, confidence was self-reported and no objective measurement of respondents' capabilities was performed. The use of Likert scales can result in central tendency bias; however, this was only reflected in some of our Likert scale responses. We feel that the responses that did reflect this bias yielded the most important results. Physician responses to clinical scenarios may not mirror behavior in an actual patient encounter. Although our sample included more diversity than other studies on physician readiness for the incorporation of genetics into clinical practice in terms of practice type (academic and non-academic) and setting (urban, suburban and rural), respondents came from one regional area of the United States.

Conclusions

Lack of experience, confidence, and resources continue to hinder

primary care physicians' optimal use of genetic testing. After more than a decade of studies of genetic tests and the PCP population, minimal progress has been made in experience and confidence. Establishing undergraduate, graduate, and continuing medical education must be a priority in order to improve and support clinician knowledge. Further research in this subject must evaluate which learning opportunities physicians would prefer. Examining the effects of age, specialty, and practice type on objective knowledge and confidence would aid in development of the right educational tools. Resources at the practice and hospital/network level including, for example, on-site genetics counseling, point of service apps on handheld devices, or EMR-based decision support, could then be tailored to primary care physicians' needs for managing the inevitable integration of genomic medicine into clinical practice.

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