Genetics and Genomics in Clinical Practice: The Views of Wisconsin Physicians

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ABSTRACT

Introduction: Decreasing costs and increased availability of genetic testing and genome sequencing mean many physicians will consider using these services over the next few years. Despite this promising future, some argue the present roadmap for translating genetics and genomics into routine clinical practice is unclear.

Objective: We conducted a pilot study to explore Wisconsin physicians' views, practices and educational desires regarding genetic and genomic testing.

Methods: Our study consists of an Internet survey (n=155) conducted in August and September 2015 and follow-up phone interviews with a portion of survey participants. Physicians of all specialties were invited to participate. Variables measured include physicians' general knowledge and experience regarding genetic and genomic testing, attitudes and perceptions toward these tests, testing intentions, and educational desires. Sociodemographic variables included gender, age, and medical specialty.

Results: In our exploratory survey of Wisconsin physicians, adult primary care providers (PCPs) lagged behind other providers in terms of familiarity and experience with genetic and genomic testing. PCPs in our sample were less likely than other physicians to feel their training in genetics and genomics is adequate. Physicians younger than 50 were more likely than older colleagues to feel their training is adequate.

Conclusions: Our exploratory study suggests a gap in physician education and understanding regarding genomic testing, which is fast becoming part of personalized medical care. Future studies with larger samples should examine ways for physicians to close this gap, with special focus on the needs of PCPs.

INTRODUCTION

Decreasing costs and increased availability of genetic testing and genome sequencing mean many physicians will consider using these tools over the next few years, with some projecting that sequencing will become fully integrated into standard medical care within 10 years.[1-5] The clinical utility of sequencing is recognized for certain diseases and in an increasing number of medical specialties,[5-6] with genetic and genomic medicine offering the promise of improved diagnostics and treatments – and patients asking physicians about the applicability of these technologies for their own care.[1,6-9] However, some experts caution the roadmap for translating genetics and genomics into routine clinical practice is unclear.[5]

Many physicians are hesitant to deepen their involvement with genetic and genomic technologies because of lack of knowledge, concerns over cost and reimbursement, and questions about clinical utility.[1,5,6,10-14] Adoption may be especially difficult for adult primary care providers (PCPs),[15] older physicians,[4] rural practitioners,[16] and specialists concerned about interpreting findings that fall outside their areas of expertise.[1] Despite these concerns, only a handful of studies have attempted to assess US physicians' experiences with genetic and genomic testing.[1,9,11,17-19] With these issues in mind, we designed a pilot survey of Wisconsin physicians exploring knowledge, experience, and attitudes regarding genetic and genomic testing, plans for using these tests in clinical practice, and perceived training needs. Respondents also were invited to participate in semistructured interviews to share additional answers and new insights. To date, there have been no similar studies that queried physicians of all medical specialties across an entire state.

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Table 1. Characteristics of the Sample

Distribution of Respondents by Key Sociodemographic Characteristics	Respondents N=155	
	n	%
Gender		
Male	89	57.4
Female	66	42.6
Age		
Under 30	5	3.2
30-39	22	14.2
40-49	32	20.6
50-59	45	29.0
60-69	41	26.5
70 or over	10	6.5
Race/Ethnicity		
White	130	83.9
Asian	12	7.8
Black/African-American	3	1.9
Hispanic/Latino	1	0.6
Did not answer/missing	9	5.8
Medical Specialty		
Adult primary care	67	43.2
Psychiatry	17	11.0
Pediatrics	10	6.4
Ob/Gyn	9	5.8
Surgery	8	5.2
Other	44	28.4

METHODS

The first part of this study consisted of an Internet survey in August and September 2015. (*Appendix "A" available at http://www.wisconsinmedicalsociety.org/_WMS/publications/wmj/pdf/116/2/McCauley__AppendixA.pdf*) E-mail invitations were sent to 12,564 Wisconsin physicians using a Wisconsin Medical Society mailing list. At the end of the survey, respondents were invited to participate in semistructured interviews. *(Appendix "B" available at http://www.wisconsinmedicalsociety.org/_WMS/publications/wmj/pdf/116/2/McCauley__AppendixB.pdf)* The study was developed by a multidisciplinary team with expertise in genetics and genomics, bioethics, law, biostatistics, and health communication. The Institutional Review Board at the Medical College of Wisconsin determined this study was exempt from oversight (PRO00024582) and formal consent from survey and interview participants was not necessary.

Data Collection and Analysis

Respondents provided sociodemographic information and medical practice characteristics via self-administered Internet questionnaires. Substantive parts of the survey used questions from previously published surveys.[8,16-19] Key *dependent variables* included questions on knowledge, training, and practice challenges phrased as dichotomous (yes/no) questions or as Likert-scale items that were transformed into yes/no responses. Physicians were asked about perceived benefits and learning needs, and to indicate their concerns about genetic and genomic testing from a list of 13 items. Those interested in training chose their desired modalities from a list of 12 items. *Independent variables* included age, medical specialty, and gender. While age was originally measured in 6 ranges, we chose to dichotomize physician responses into 2 categories: "younger than 50" and "50 or older." Owing to the modest size of our sample, we reduced all specialties into 6 categories: Adult Primary Care (family medicine, internal medicine, geriatrics), Psychiatry, Pediatrics, Ob/Gyn, Surgery (general surgery, neurosurgery, other surgery), and "Other." For heuristic purposes, we further reduced the data by categorizing physician specialties as either "Adult Primary Care" or "Other" specialty.

Statistical analysis was performed with IBM SPSS Statistics for Windows, Version 22.0.[20] Our sample size precluded the possibility of using inferential statistics; thus, we ran crosstabular analyses to identify associations between all dependent and independent variables. Physicians who agreed to participate in semistructured interviews were asked 12 questions that paralleled the structure of the survey in order to provide detailed examples of physician views. Interview times varied, with an average duration of 18 minutes. Transcripts were analyzed with a 3-stage qualitative analysis process.[21] During structural coding, the principal analyst coded textual elements in each transcript corresponding with answers to the substantive questions in our interview schedule. This was enhanced by the use of QSR NVivo 10, an ethnographic data management software program. [22] This stage was followed by immersion/crystallization,[23] which involves immersing deeply in key portions of coded data and then backing away at regular intervals for reflection and second-level theme formation. The lead author performed these analytical procedures and generated NVivo output reports for cross-checking by 2 other coders. All authors reviewed the results of these processes and contributed to the summary of qualitative findings. In this study, we adhered to best practices for conducting and presenting mixed-methods research.[24]

RESULTS

Sample Characteristics

One hundred fifty-five physicians completed our online survey; their key sociodemographic and practice characteristics are reported in Table 1. More than half were men, yet the percentage of women in the sample is greater than contemporary estimates for Wisconsin physicians.[25-27] The modal age range for physicians in our sample was 50 to 59 years (29%), compared to 45 to 54 years (23%-29%) in 2 recent statewide population-based samples.[25-26] In terms of medical specialty, we slightly oversampled with respect to adult PCPs and psychiatrists. Regarding race and ethnicity, the white/nonwhite ratio in our sample is similar to that of physicians statewide.[25-27] Finally, our survey respondents were distributed evenly across urban, suburban, and rural practice settings, representing an oversampling of physicians from suburban

and rural settings. Nineteen physicians participated in semistructured interviews; they included 10 women and 9 men with a mean age of 53. Most interview participants were white, with 14 coming from urban or suburban practice settings. Six were psychiatrists, five were PCPs, and the remainder represented other specialties.

General Knowledge and Experience

Our survey results suggest physician specialty and age may be the primary drivers of key outcome variables (See Table 2). The size of our survey sample prevented us from making detailed comparisons between physicians from many specialties and subspecialties, however, PCPs in our sample were much less likely than other physicians to say they are familiar with genetic and genomic testing. In contrast, two-thirds of ob/gyn specialists and more than one-third of psychiatrists said they are familiar. PCPs in our sample were also less likely to feel adequately informed about the availability and clinical utility of these tests, while more than half of responding pediatricians said they are adequately informed. Younger physicians were more likely than colleagues 50 or older to have received formal training in genetic/genomic medicine. Finally, about 30% of physicians with practices in urban or suburban settings felt adequately informed about availability and clinical use, compared to 14% in rural settings.

The physicians interviewed generally spoke about the limited use of genetics and genomics in their own practices. However, some spoke about the promise of pharmacogenomics for fine-tuning psychiatric medications. (See Table 3 for a summary of other interview results.) While respondents said many patients are not yet asking about genetic and genomic tests, some physicians reported a heightened sense of interest in oncology applications. For example, a 44-year-old female breast surgical oncologist in suburban practice said:

> *"I see a fair amount of breast cancer patients, as well as patients who come seeking medical attention in terms of risk assessment and strategies for risk reduction for breast cancer. So, if they fit the NCCN guidelines to consider genetic testing or counseling, then that gets offered in my office."*

Some physicians said patients are asking about implications of genetic/genomic tests for prenatal concerns or as an extension of family medical history. Finally, most said the ability to use genetic and genomic testing is at least an important *consideration* in their practices, with special emphasis on select patients. Among physicians who said genetic and genomic testing is not currently important in their practice, some said testing may become important in time.

Table 2. Summary of Selected Survey Results

	Respondents	
	n	%
General Knowledge and Experience Regarding Genetic and Genomic Testing		
I am familiar with genetic/genomic testing		
Adult Primary Care Physicians	6	9.0
All Other Physicians	23	26.1
≥ 50 years old	19	19.8
Under 50	10	16.9
I have had some type of formal training in genetic/genomic medicine.		
Adult primary care physicians	18	26.9
All other physicians	30	34.1
≥ 50 years old	25	26.0
Under 50	23	39.0
I feel adequately informed about the availability and clinical applications of genetic/genomic testing		
Adult primary care physicians	8	11.9
All other physicians	30	34.1
≥ 50 years old	21	21.9
Under 50	17	28.8
Attitudes Toward Genetic Testing and Genome Sequencing		
I have sufficient knowledge to counsel patients about genetic risk for disease		
Adult primary care physicians	16	23.9
All other physicians	29	33.0
≥ 50 years old	24	25.0
Under 50	21	35.6
Testing Intentions		
I anticipate ordering/recommending a genetic/genomic test within the next 6 months		
Adult primary care physicians	29	43.3
All other physicians	44	50.0
≥ 50 years old	42	43.8
Under 50	31	52.5
Educational Desires		
I feel that my professional training in genetics/genomics is adequate		
Adult primary care physicians	2	3.0
All other physicians	19	21.6
≥ 50 years old	10	10.4
Under 50	II	18.6
I would be interested in further professional education in genetics/genomics		
Adult primary care physicians	54	80.6
All other physicians	69	78.4
≥ 50 years old	79	82.3
Under 50	44	74.6

Note: For brevity, results that pertain to specific specialties and other physician characteristics have been omitted.

Attitudes Toward Genetic Testing and Genome Sequencing

Between one-half and two-thirds of physicians in our survey sample said there are now sufficient benefits to warrant *genetic* testing for determining cancer type, prognosis, and/or targeted treatment; diagnosis of Mendelian or rare diseases; reproduction and family planning; and identifying genetic risk factors for adult-onset complex diseases. Nearly 55% said there are now sufficient benefits to warrant *genomic* testing for determining cancer type, prognosis, and/or targeted treatment. Most respondents said it is important for them to learn about a variety of new advances in genetic testing, with emphasis on determining drug and dose compatibility for a patient, and diagnosing and identifying genetic risk factors for adult-onset complex diseases. A slightly smaller major-

Table 3. Summary of Other Representative Interview Results

	Themes	Examples
General Knowledge and Experience	Limited exposure	"I don't have much experience; internists don't get much genetics/genomics training."
	What patients ask	"My patients ask about the risk of breast, ovarian or primary peritoneal cancer."
	Importance of genetic/genomic medicine	"Testing is important for certain patients regarding cardiac conditions or cancer."
Attitudes Toward Genetic Testing and Genome Sequencing	Personalized medicine is promising	"I'm interested in screening patients because my family faces certain genetic risks."
	Costs and benefits: clinical utility	"Let's get the right test to the right patients and explain the consequences."
	Concern over incidental findings	"If you test willy-nilly, you'll get noise. And noise leads to poor treatment."
Testing Intentions	Timely results for a reasonable price	"I'd like good and quick results to help patients better metabolize pain meds."
	A premium on tests that come with clear guidance	"Parents of children with birth defects need sound guidance about future pregnancies."
	Insurance companies sometimes put up harriers	"It's tough for me to order when insurance won't pay for tests or genetic counseling."
Educational Desires	General enthusiasm to learn more	"We have huge potential for impacting patients' lives by learning their genetic quirks."
	Self-directed online courses are best	"I don't have much time. But in the past, I did a ton of online CME during night shifts."
	Despite interest, there is precious little time to learn	"How much time does it take to become minimally proficient with this kind of testing?"

ity of respondents said it is important for them to learn about the same advances in *genomic* testing. Regarding testing concerns, about 70% of physicians worried that patients may interpret test results incorrectly. A smaller number were concerned that test results could lead to discrimination by insurers or that the validity or accuracy of results is questionable. Twenty-four percent of PCPs said they had sufficient knowledge to counsel patients about genetic disease risk, while one-third of all other physicians felt similarly (Table 2). Only about 20% of respondents said they had sufficient time in their practices to counsel patients about genetic risk for disease.

Most physicians interviewed found the prospect of personalized medicine promising, while some found it to be complex, citing a need to know which tests are applicable to their patients and may help to improve patient outcomes. Many called for the development of more contextual information about genetic testing and genome sequencing – actionable, evidence-based guidance formatted into easy-to-use decision aids. A fair number of physicians voiced concerns about insurance coverage and overall affordability. Others, especially psychiatrists, said the practice of pharmacogenomics holds great promise for patients who fail to respond to early medication trials.

Interviewees raised a variety of concerns regarding the clinical utility of genetic and genomic testing, including affordability and access, discrimination by insurers, and the possibility patients will misinterpret test results. Some of the most interesting comments concerned the handling of incidental findings, including this one by a 54-year-old male hematology and oncology specialist in urban practice.

> *"[Depending on the] particular panels of genes, you certainly get a lot of information that you are not sure what to do with. We find out mutations in all kinds of genes that, right now, aren't actionable, given [that] the quality of the data and understanding what they mean is not so clear. In that setting you can develop a bias of over-treatment based on perceived risk that may not necessarily be well vetted from a research standpoint."*

One psychiatrist also noted some patients learn things about themselves they did not want to know, prompting the need for psychoanalytic investigation into fears about illnesses they might develop later in life.

Testing Intentions

About 43% of PCPs in our survey anticipated ordering genetic or genomic tests within the next 6 months (Table 2), while roughly 60% to 75% of ob/gyn specialists, surgeons, pediatricians and psychiatrists said they would. Almost 44% of respondents said genetic and genomic tests are not applicable in their practice, while nearly 28% said they do not have enough knowledge about these tests.

Equal numbers of interview respondents said their level of ordering likely would stay the same in the near future or would increase if tests are shown to be efficient and cost effective. Most expressed a desire for clear guidance regarding the scientific reliability and clinical applicability of these tests. Others wanted timely and relevant results that suggest concrete solutions, including a 45-year-old female family physician in a rural practice who pondered the results of genomic testing aimed at uncovering causes and treatments regarding her own disease:

> *"You know, there may not always be a simple solution, but there are some things where there's clinical applicability and relatively simple nutritional solutions to get around these little SNPs. So, I really think this will be the wave of the future."*

Participants also spoke about several barriers to ordering genetic/genomic tests including high cost, insurance coverage, physicians' own lack of knowledge and experience, and the time commitment involved with ordering, interpreting, and counseling patients.

Educational Desires

Nearly three-quarters of survey respondents said their professional genetics/genomics training is inadequate, with PCPs being much more likely to feel this way. Physicians younger than 50 were more likely than older colleagues to feel their training is adequate (Table 2). Nearly 80% of respondents said they would be interested in further education in genetics/genomics; 88% of physicians from rural practices felt this way, compared to 75% of physicians in urban or suburban settings. More than 70% of respondents said they would be willing to devote time to continuing medical education (CME), with more than 60% preferring to receive additional training through self-directed online courses and 53% through in-person CME. Respondents also stated preferences for education via professional meetings (45.2%), journal publications (38.7%), and grand rounds or other in-house seminars (38.7%).

Most physicians we interviewed expressed unqualified enthusiasm for further education about genetic testing and genome sequencing. Some respondents, including younger physicians, noted the lack of genetics training during medical school. One 34-year-old male family physician practicing in a rural practice setting said:

> *"There was the requisite preclinical course on genetics, which was essentially a unit within the larger course on biochemistry. That was, frankly, a fairly cursory review compared to the deeper dive into genetics I had as an undergrad. This was very basic stuff; it did not get into things like whole genome sequencing. It really talked a lot more about specific case presentations of genetic disorder as opposed to some of the testing that would go along with it. So, more 'Here's what it looks like' [and] less about 'Here's how to find it.'*

Other participants spoke of PCPs' need for more in-depth training. Respondents also listed a few barriers to additional training, mainly regarding the lack of time physicians have within the context of a busy clinical practice. Some spoke of a lack of motivation to engage in such training given competing CME, and others acknowledged a lack of basic understanding or awareness regarding genetic/genomic testing on their part and amongst their colleagues. Finally, some respondents lamented the paucity of basic educational programs that would enable physicians who are not genetic specialists to become proficient enough to utilize certain tests in their own practice.

DISCUSSION

This exploratory study summarizes the views of a small sample of Wisconsin physicians about genetic and genomic testing, with an emphasis on general knowledge and experiences, attitudes toward testing, testing intentions, and educational needs. Consistent with current literature,[1,9,11,17,18] our study found that while physicians increasingly see the value of these tests, relatively few have significant experience with them or feel prepared to use them. Perhaps the most significant finding is that adult PCPs lagged behind other physicians in each of these areas and were less likely to feel their training in genetics/genomics is adequate. There are many potential explanations for these findings; regardless, this knowledge gap amongst PCPs is important to address for several reasons. For example, given the sheer volume of patients seen, PCPs likely serve a greater number and variety of people who may benefit from genetic/genomic testing than other specialists. Also, PCPs have been proposed as potential surrogates for genetic counselors, who are too few in number relative to the demand for their services.

Our study also suggests that younger physicians are more likely than older colleagues to report having formal training in genetics/genomics, and to feel their training is adequate. This finding should be interpreted with caution since self-reported genetic and genomic knowledge does not always correlate with the level of knowledge that physicians actually possess. Future studies could utilize exams evaluating participants' genetics/genomics knowledge and compare the results with self-reported knowledge. Furthermore, confidence in genetic/genomic competency must be tempered by the fact that the rapid pace of new developments in these areas may quickly render anyone's present knowledge obsolete. Thus, medical educators should continue to refine genetics/genomics curricula in medical school and residency training, and develop effective CME to help practicing physicians stay up-to-date on technologies applicable for their patients. Finally, survey respondents who practice in rural settings were about half as likely as physicians from urban/suburban settings to feel adequately informed about genetic/genomic medicine. Fortunately, these physicians recognize their knowledge deficits and were more likely to express interest in further education. This may be especially important in states like Wisconsin, where barriers associated with cost and lengthier wait times for testing and results may prevent patients in rural areas from realizing the full benefits of genetic/genomic technologies.[16]

This study is not without limitations. First, the sample size is small. However, we can make statements with confidence about key questions that pertain to differences between physicians from 2 age groups and 2 specialty categories of "Adult Primary Care" and "Other." Though our sample consists of 155 physicians from a variety of specialties, the number of physicians from many specialties was too small to deliver sufficient power for the use of inferential statistics. Finally, our sample may be biased toward physicians with a preexisting interest in genetics/genomics. Despite these shortcomings, our study offers an early look at the differences between primary care and other specialist physicians in Wisconsin regarding several key questions that pertain to their experience with this rapidly advancing field. Future studies with larger state-

wide samples might use our survey and interview questions to tease out additional details.

Many foresee rapid advances in genetic testing and genome sequencing over the next decade, with inevitable implementation into clinical practice. Our study adds to a small but growing body of literature documenting the growing pains of genetic and genomic medicine. Now is the time to ensure that knowledge about these technologies—and their importance to personalized medicine—is shared widely among physicians. To further deploy these technologies for optimal health outcomes at the population level, medical educators need to move the use of genetics/genomics beyond the realm of early-adopting physicians and into the hands of those who serve more diverse populations, including groups that are now underserved by our health care system.[5]

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