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Texas Physicians' Awareness and Utilization of Genetic Services

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TEXAS PHYSICIANS' AWARENESS AND UTILIZATION OF GENETIC SERVICES

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A

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The number of disorders for which genetic testing is available has increased nearly 500% in the past 15 years. Access to the majority of genetic tests and services hinges on physicians' ability to identify patients at risk for genetic disease and provide appropriate testing and counseling or refer to genetic specialists. Recent research demonstrates the need for referrals to genetic specialists by showing that many physicians lack skills required to perform appropriate genetic services, such as making proper risk assessments, providing genetic counseling, ordering genetic testing and interpreting results. However, little research exists on physicians' awareness and utilization of genetic services. In this study, an electronic survey evaluating practicing physicians' awareness of, utilization of and perceived barriers to genetic services in Texas was distributed via state physician organizations. Of the 157 participants, approximately half reported they were moderately or very aware of genetic testing and services in their area. Very few reported awareness of telemedicine services. Over two-thirds reported never or rarely referring to genetic counselors or other genetic specialists, despite 75% reporting they had noticed an increased impact of genetics on their field and 61% reporting they had discussed genetics more in their day-to-day practice in the last 5-10 years. Only 20% reported genetics was very integral to their specialty. Over three-fourths of all participants indicated interest in learning more about genetics, genetic testing and genetic services. Among the most frequently chosen barriers to genetic counselors were awareness-related barriers such as not knowing how to refer to a genetic counselor. Responses to many items varied significantly by medical specialty. The results identify a need to increase awareness of genetic services and referral logistics. Specific findings can help direct outreach efforts to educate clinicians, such as developing clinically meaningful, specialty-specific educational objectives.

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Background

Sixty years have passed since the discovery of the structure of DNA and a quarter of a century since the commencement of the Human Genome Project. Knowledge of the composition of our genetic code unsurprisingly prompted massive research efforts to link genes to disease and develop methods for clinical genetic testing. The availability, affordability and accessibility of genetic testing services continue to expand for a constantly growing menu of genes relevant to a number of medical specialties. Over 2,900 genes related to human disease have been identified, and over 17,000 genetic tests for more than 4,300 human conditions are commercially available (Rubinstein et al., 2014). Accordingly, a substantial number of today's patients would benefit from genetic services such as genetic evaluation, counseling and testing.

Diagnosis of a genetic condition can provide relief to families by identifying a reason for recurring health issues in the family history or developmental differences persisting in a child. For many, a documented genetic etiology for health or intellectual issues decreases barriers to medical care, functional therapies and other services. It may allow an individual to undergo surveillance for additional medical risks associated with the condition, which may save or drastically improve the quality of a life. Identification of a genetic mutation in one family member also introduces the option of predictive testing for other relatives, which may influence personal and reproductive choices.

Since physicians typically serve as patients' primary or only source of healthcare, patients rely on them for medical information and, ideally, trust that their physician grants them the highest quality of care (Miller et al., 2010). The complexity of genetic testing options and result interpretation, in combination with inadequate provider knowledge, can lead to deficient patient care. High-quality care will increasingly include access to genetic services, and physicians embody a vital link between patients and these services. Unfortunately, research shows physicians are generally ill-equipped to provide genetic services.

A fundamental requirement for providing genetic services is a basic understanding of genetic concepts and conditions. Previous research shows that on average, physicians lack this knowledge. For

example, Baars, Henneman, and Ten Kate (2005) found that the majority of general practitioners evaluated lack basic genetics knowledge such as understanding genetics-related terms. In 2013, 73.7% of physicians surveyed by Klitzman et al. rated their knowledge of genetics as "very/somewhat poor." In other studies, physicians recognize their genetics illiteracy and report that it causes low confidence in handling genetics-related issues; they admit to relying on other providers to recognize patients in need of a genetics evaluation and to ensure this service is rendered (Houwink et al., 2011; Metcalfe, Hurworth, Newstead & Robins, 2002; Nippert et al., 2011). Without knowledge that certain genetic conditions and testing exist, physicians are incapable of promoting the best health care for the patient and family members.

Risk assessment skills also underpin the provision of genetic services. Several studies emphasize physicians' inability to properly perform a risk assessment to identify individuals at risk for genetic conditions based on personal and family history (Baldwin et al., 2014; Bellcross et al., 2011; Bonham, Jenkins, Stevens & McBride, 2008; Pompilii et al., 2014; Trivers et al., 2011). In a vignette-based study, only 41% of 979 physician participants indicated they would recommend a referral for genetic services for women at high-risk of ovarian cancer (Trivers et al., 2011). Trivers et al. (2011) and Bonham et al. (2008) both reported physicians may over-refer low risk patients to genetic counselors and under-refer high risk patients. In 2014, Leandro, Paneque, Sequeiros and Porto conducted a study on genetic counseling referrals for patients at risk for hereditary hemochromatosis. They found physicians lacked the knowledge necessary to assess patients' risk for this condition and emphasized the exigency of a genetics evaluation for early detection and treatment of the disease (Leandro et al., 2014). In a study focused on prenatal patients, Pompilii et al. (2014) found 6.4% of the patients referred to genetic counseling for advanced maternal age alone had at least one additional genetic risk factor that surfaced during the counseling appointment, the majority (72.3%) of which were consanguinity, Mendelian disorders or chromosome conditions. The patients' obstetricians or other referring doctors had not accurately assessed potential reproductive risks. Across several medical specialties, inability to recognize patients at increased risk precludes entire families from gathering meaningful genetic information.

Physicians similarly lack knowledge of genetic testing, another central requirement for providing genetic services. Over half of the physicians in a large scale study reported they were not knowledgeable about genetic tests and 43% reported they were only somewhat knowledgeable (Mainous, Johnson, Chirina & Baker, 2013). In conjunction with physicians' lack of knowledge about genetics in general, lack of knowledge about genetic testing can lead to ordering the incorrect genetic test. Common pitfalls include choosing tests targeting the incorrect condition or gene, ordering panels that are not sufficiently comprehensive or designating the wrong testing technology. Careful selection of the most appropriate testing candidate in a family is also essential since testing certain family members may yield uninformative results. All of these issues cause failure to deliver sufficient genetic information to patients and wasting of healthcare dollars. In fact, one study showed when genetic counselors reviewed test orders at a genetics laboratory, referring institutions saved an average of $48,000 per month by suggesting more appropriate testing strategies (Miller et al., 2014). Interpretation of genetic testing results increasingly requires some background in genetics, therefore many physicians are at risk of misunderstanding and misrepresenting results (Brierley et al., 2010)

Adding further complexity, genetics issues can be laden with profound ethical, psychosocial and legal complexities, and physicians' unfamiliarity with these topics can lead to cases of psychological harm and unethical practices (Demmer, O'Neill, Roberts & Clay, 2000; Michigan Association of Genetic Counselors, Inc., 2012). Bensend, Veach and Niendorf (2014) cited fifteen cases of adverse emotional effects on patients who received genetics services from non-genetics professionals in Minnesota. Klitzman et al. (2013) found 87% of internists surveyed reported very or somewhat poor knowledge of genetic testing guidelines, and 80% saw a need for more training on how to protect the privacy of their patients' genetic information. Lowstuter et al. (2008) found that of the 1,181 physician, nurse practitioners and other medical association members sampled, 75% thought patients would decline genetic testing if offered due to fear of genetic discrimination, and over 60% were unaware of state and federal laws regarding genetic discrimination by health insurance companies. They demonstrated that the more a participant viewed genetic discrimination as a threat to patients, the less likely the participant was

to refer a patient to genetic services, showing that misunderstanding or unawareness of relevant laws can obstruct patient access to genetic services (Lowstuter et al., 2008).

As demonstrated by prior research, many physicians lack at least some essential skills to provide thorough genetic services including recognizing when to refer patients to genetic services. Given already oversaturated medical school curriculum and the tremendous number of genetic conditions and corresponding tests, blame cannot rest on physicians for this gap in knowledge. The data simply highlight that quality genetics services necessitates an expert with specialized training in genetics, just as is the case for other medical specialties. Genetic experts including MD and PhD geneticists, genetic counselors, genetic nurses and other associates possess the skills needed for responsible dissemination of genetic services. Genetic counselors in particular receive comprehensive training on the medical, molecular, hereditary, psychosocial, ethical, legal, logistical and other elements of genetic conditions and testing. Genetic counselors are not only qualified to guide and counsel patients through these intricacies, they also tend to have more time to thoroughly discuss such issues (Gottschalk & Flocke, 2005; Centers for Disease Control & Prevention, 2013; National Society of Genetic Counselors, 2014).

In other words, physicians are not expected to have the depth of knowledge, skills or time to provide complete genetic services themselves, but they still play a critical role in patient access to services by facilitating the overwhelming majority of referrals to genetic counselors and other genetic specialists. Fulfilling this role requires the ability to properly screen and refer patients. Foremost, physicians must perform adequate genetic risk assessments. Educational campaigns should target this area, since previous research has identified the need. Second, physicians must be informed about services and benefits genetic counselors provide, and finally, they must know how to refer to these services logistically.

As past studies have addressed knowledge of basic genetics concepts, risk assessment skills and ethical awareness, the present study focuses on physicians' awareness of genetic testing and services available to their patients as well as frequency of referral to genetic specialists. The study also aims to capture perceived barriers to genetic services and physician interest in learning more about genetics and

genetic services. Multiple physician specialties were pertinent to the current research since genetic testing increasingly encompasses a wide array of rare and common conditions across many specialties. Obstetricians, oncologists and pediatricians were of particular interest since these areas traditionally predominate the genetic counseling field. Family and internal medicine physicians were also targeted due to their role as primary care physicians. Multiple other medical specialties were included since genetic conditions present with various clinical findings and currently available genetic testing is relevant to a number of fields.

Results of the study will determine how providers currently utilize genetic services in Texas and will validate the need to educate clinicians about genetic needs and services in the state. Together, these findings can conceivably improve patient care by increasing appropriate genetics referrals and improving access to genetic services.

Methods

Instrumentation

A 39-item survey was developed targeting several overarching domains: demographic information about the participant, demographic information about the participant's medical practice, general perception of genetics in medicine, awareness of genetic services, utilization of genetic services, interest in learning about genetics and barriers to genetic services. The survey was administered using the electronic survey tool, REDCap v.5.9.11 (https://redcap.uth.tmc.edu/index.php?).

Participants and Procedures

Currently practicing physicians in Texas were eligible to participate in this study, which was approved by the Institutional Review Board at the University of Texas Health Science Center at Houston. Participants were recruited via Texas medical organizations. Using publically available contact information, we contacted 16 county medical societies, 14 state specialty societies, 5 large physician groups and 5 other physicians groups by email or phone with a request to distribute an electronic survey link to members. Five county medical societies, 5 state specialty organizations, 2 large physicians groups and one other organization agreed to distribute the link internally. Ten organizations sent the link within a routine e-newsletter, and 3 sent the link by direct email blast. Over a 17-week period, there were 162 total survey participants. Five of the respondents were excluded after reporting they were not currently practicing, resulting in 157 participants. Response rate could not be calculated due to unavailability of the total number of physicians who received an invitation to participate in the survey.

Data Analysis

Categorical data was compared across groups using contingency tests (Chi-square or Fisher exact test). Spearman's rank correlation tests were used to evaluate correlations between different Likert scales. All analysis was performed using Stata (v. 13, College Station, TX). Statistical significance was assumed at a Type I error rate of 5%.

Results

Demographics

Participant demographic characteristics and specialty of practice are displayed in Table I along with selected available demographic information on the target population, which included all physicians practicing in Texas.

Table I Study Population and Known Target Population Demographics

	Study Population	Target Population[1]
Sample size (n)	157	56,210
Gender		
Male	61%	68%
Female	39%	32%
Age		
24-30	3%	2%
31-40	30%	25%
41-50	26%	28%
51-60	21%	23%
61-70	16%	15%
71-80	4%	6%
Years practicing medicine		
0-5	19%	
6-10	17%	
11-15	12%	
16-20	13%	
21-25	11%	
26+	28%	
Practice Setting		
University Medical Center	34%	
Private Hospital/Medical Facility	7%	
Public Hospital/Medical Facility	7%	
Physician's Private Practice	25%	
Private Practice-Self-Employed	23%	
Other	4%	
Practice Location		
Urban	56%	
Suburban	31%	
Rural	12%	
Other	1%	
Specialty		
Dermatology	32%	
Ob/Gyn	17%	
Family Medicine	12%	
Internal Medicine	10%	
Other	11%	
Pediatrics	7%	
Surgery	6%	
Neurology	5%	

[1]Texas Medical Board (2015)

General Perception of Genetics

Overall, 42% (66/157) of participants felt genetics was moderately or very integral to patient care in their specialty of practice. However, these proportions varied by specialty (Table II) ($p<0.001$). Participants who reported that genetics was not integral to their practice were more likely to be self-employed in a private setting (42%, 15/36, $p=0.005$) compared to physicians who reported that genetics was at least moderately integral (15%, 10/66). No significant differences were found between gender, age, number of years practiced or practice location for this measure.

A high percentage (73%, 115/157) of participants reported they had noticed an increase in the impact of genetics on their field in the last 5-10 years. There was significant variation according to specialty (Figure 1) ($p=0.047$) and age. A higher proportion of participants age 61-80 (31%, 10/32) reported they had not noticed an increased impact compared to younger participants age 31-60 (12%, 15/121, $p=0.014$).

There was a moderate correlation between whether participants had noticed an increased impact of genetics in the last 5-10 years and perceived integrality of genetics to patient care in his/her specialty (Spearman's rho=0.38, $p<0.001$). Nearly all the participants who felt that genetics was very integral reported noticing an increased impact compared to just over half of those who felt that genetics was not at all integral (97%, 30/31 vs. 53%, 19/36). Among participants who reported an increased impact, over half had noticed it in articles in medical journals (58%, 91/157) or in presentations at medical conferences (57%, 90/157). Forty-two percent (66/157) of participants reported noticing the increased impact from their patients.

Overall, half (50%, 79/157) of the participants reported they had discussed genetics and/or genetic testing more in their day-to-day practice in the last 5-10 years, and there was significant variation between specialty as illustrated in Figure 1 ($p<0.001$).

Figure 1 highlights variation across selected specialties between the proportions of participants who perceived genetics as moderately or very integral to their specialty, indicated that they had noticed an increased impact of genetics on their field in last 5-10 years and indicated that they had discussed genetics more in their day-to-day practice in the last 5-10 years. As illustrated, Ob/Gyns and

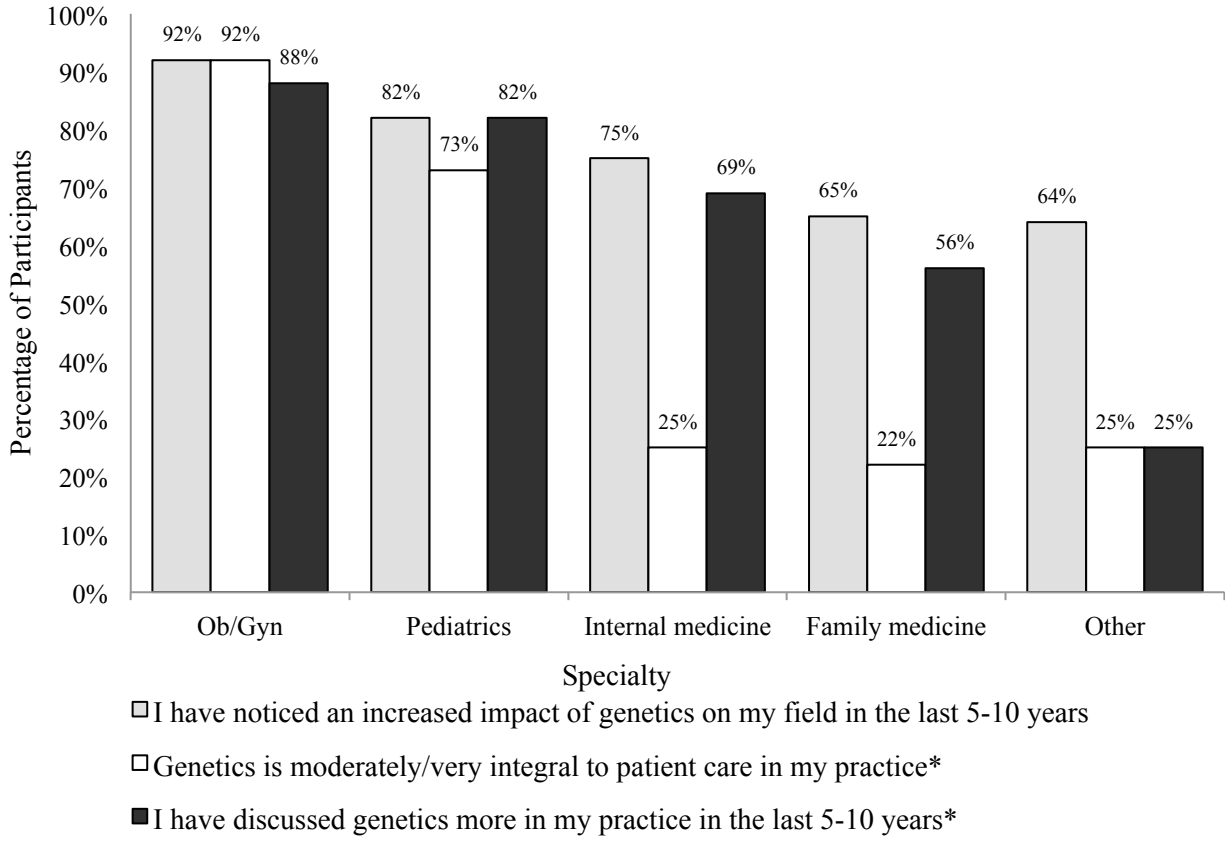
Table II Responses to various questions according to specialty

Specialty		Response, % (n)			
		Not at all	Slightly	Moderately	Very
Pediatrics	How integral is patient care in your specialty of practice?*	-	27 (3)	36 (4)	36 (4)
Ob/Gyn		-	8 (2)	31 (8)	62 (16)
Neurology		25 (2)	13 (1)	25 (2)	38 (3)
Family Medicine		35 (6)	41 (7)	18 (3)	6 (1)
Internal Medicine		27 (4)	47 (7)	13 (2)	13 (2)
Dermatology		22 (11)	52 (26)	20 (10)	6 (3)
Other		47 (8)	18 (3)	24 (4)	12 (2)
Surgery		50 (5)	30 (3)	20 (2)	-
Pediatrics	How familiar are you with the roles a genetic counselor can provide?*	-	18 (2)	27 (3)	55 (6)
Ob/Gyn		4 (1)	8 (2)	31 (8)	58 (15)
Neurology		13 (1)	25 (2)	13 (1)	50 (4)
Family Medicine		6 (1)	33 (6)	44 (8)	17 (3)
Internal Medicine		6 (1)	31 (5)	38 (6)	25 (4)
Dermatology		8 (4)	43 (22)	33 (17)	16 (8)
Other		29 (5)	24 (4)	29 (5)	18 (3)
Surgery		40 (4)	30 (3)	30 (3)	-
Pediatrics	Are you aware of the available genetic services in your area?*	-	9 (1)	55 (6)	36 (4)
Ob/Gyn		8 (2)	15 (4)	23 (6)	54 (14)
Neurology		13 (1)	25 (2)	13 (1)	50 (4)
Family Medicine		39 (7)	28 (5)	22 (4)	11 (2)
Internal Medicine		31 (5)	13 (2)	38 (6)	19 (3)
Dermatology		41 (21)	27 (14)	22 (11)	10 (5)
Other		41 (7)	24 (4)	24 (4)	12 (2)
Surgery		67 (6)	11 (1)	22 (2)	-
Pediatrics	Are you aware of genetic testing available to your patients?*	-	18 (2)	45 (5)	36 (4)
Ob/Gyn		-	4 (1)	24 (6)	72 (18)
Neurology		25 (2)	13 (1)	13 (1)	50 (4)
Family Medicine		17 (3)	44 (8)	28 (5)	11 (2)
Internal Medicine		31 (5)	19 (3)	31 (5)	19 (3)
Dermatology		22 (11)	49 (25)	16 (8)	14 (7)
Other		41 (7)	12 (2)	35 (6)	12 (2)
Surgery		56 (5)	22 (2)	22 (2)	-

*$p<0.05$

Notes: A dash (-) signifies "0 (0)"

Figure 1 Variation between proportions of participants in certain specialties who selected particular responses



*p<0.05
Note: Specialty groups neurology, dermatology and surgery are not displayed in this figure.

pediatricians had roughly concordant proportions between the three responses, with comparable percentages in each category. Family and internal medicine physicians were more likely to report noticing an increased impact and discussing genetics more than they were to report that genetics was at least moderately integral to their specialty. Similarly, the "other" specialties were less likely to feel that genetics is integral or claim that they have discussed genetics more, yet a high proportion of those participants reported noticing an increased impact of genetics on their field.

Awareness of Genetic Services

A slightly higher percentage of participants reported being moderately or very familiar with the roles a genetic counselor can provide (60%, 94/157) compared to moderately or very aware of genetic testing (50%, 78/157) and genetic services in their area (47%, 74/157). Eleven percent (17/157) reported

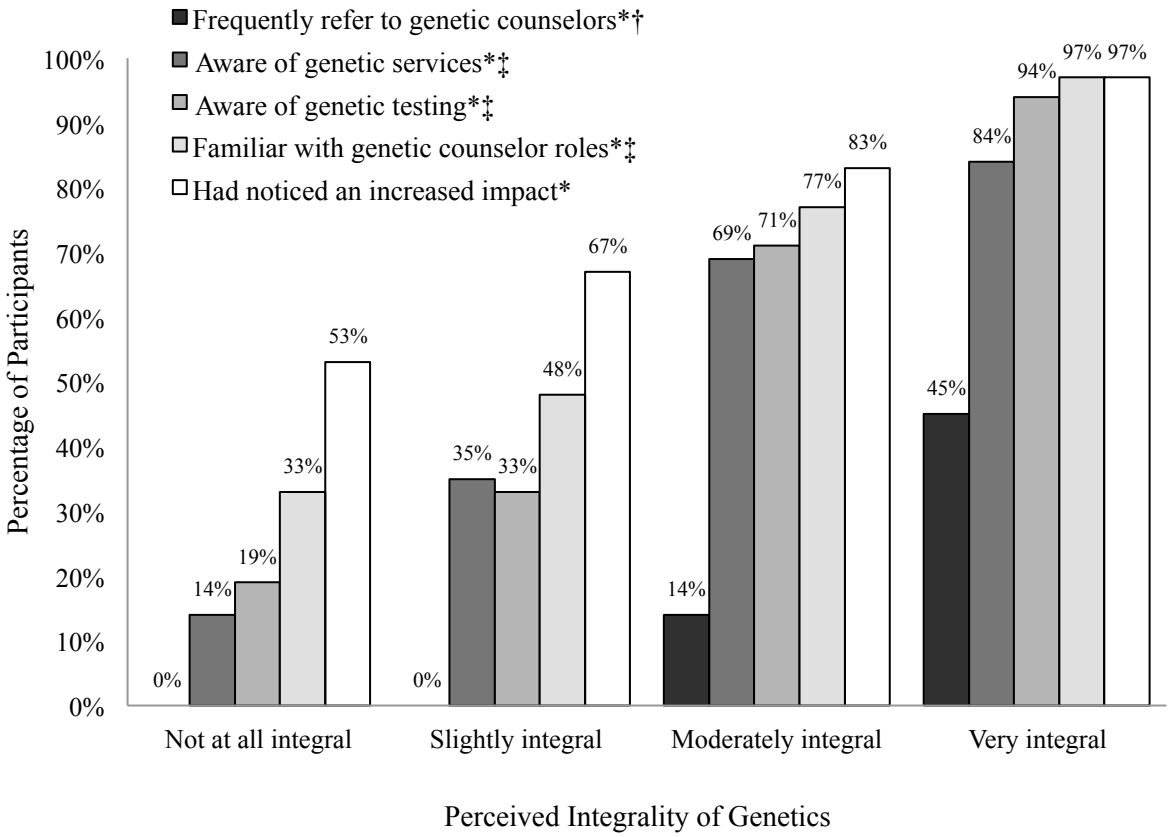
they were not familiar with roles a genetic counselor can provide, 21% (33/157) reported they were not aware of genetic testing and 31% (49/157) reported they were not aware of genetic services in their area. Participants were least aware of telemedicine services, with 80% (125/157) reporting they were not aware, 12% (18/157) reporting they were slightly aware and only 7% (11/157) reporting they were moderately or very aware of this service. More than half of participants (53%, 83/157) reported knowing how to refer patients to genetic services, and just under half (49%, 77/157) reported knowing of a specific laboratory that performs genetic testing that they would use.

Familiarity with genetic counselor roles, awareness of genetic testing and awareness of genetic services varied by specialty (each measure with $p<0.001$). Ob/Gyns consistently had the highest percentage reporting being very familiar or very aware. Awareness of services in the area also varied by practice location ($p=0.024$), with 57% (50/88) of urban participants reporting they were moderately or very aware of services in their area and only 35% (17/49) of suburban participants and 32% (6/19) of rural participants reporting the same. There was no significant difference between gender or practice setting for any of these measures, between age or practice location for familiarity with genetic counselor roles and awareness of genetic testing, or between number of years practicing for awareness of testing.

There was a moderately high correlation between perceived integrality of genetics and awareness of genetic services in the area (Figure 2) (Spearman's rho=0.58, $p<0.001$). Over two-thirds (68%, 21/31) of the participants who felt that genetics was very integral were very aware of services in their area, whereas almost two-thirds (64%, 23/36) who felt that genetics was not at all integral were not aware of services. Likewise, 90% (28/31) of those who felt genetics was very integral reported knowing how to refer to genetic services, whereas 75% (27/36) of the participants who felt that genetics was not at all integral reported not knowing how to refer to genetic services. Similar correlations were found between perceived integrality of genetics and familiarity with genetic counselor roles, awareness of genetic testing and knowledge of a laboratory that provides genetic testing (each measure with $p<0.001$).

Twenty-four percent (16/66) of those who felt genetics was moderately or very integral to their practice reported they were not aware or were slightly aware of genetic services in their area. Of these

Figure 2 Relationship between perceived integrality of genetics and selected awareness and utilization measures



†Bars are limited to participants who reported "somewhat frequently" or "very frequently"
‡Bars are limited to participants who reported "moderately aware" or "very aware"
* Significant trend ($p<0.001$)

participants, 56% (9/16) were age 40 or younger and 44% (7/16) had been practicing for 0-5 years. Thirty-eight percent (6/16) of these participants were dermatologists and 25% (4/16) were Ob/Gyns.

The majority of participants were interested in learning more about genetics and genetic testing relevant to their field (77%, 121/157) as well as genetics services available to their patients (78%, 123/157). However, interest in learning about genetics and genetic testing varied by specialty (p=0.018), with 100% (8/8) of neurologists, 91% (10/11) of pediatricians, 84% (43/51) of dermatologists, 78% (14/18) of family medicine physicians, 73% (19/26) of Ob/Gyns, 69% (11/16) of internal medicine physicians and 40% (4/10) of surgeons reporting they were interested in learning more. There was no significant difference between specialties for interest in learning about genetic services. Participants who

were interested in learning more about genetics, genetic testing and genetic services were more likely to have noticed an increase in the impact of genetics on their field than those who were not interested, with 78% (94/121, p=0.026) of those interested in learning about genetics and genetic testing and 79% (97/123, p=0.004) of those interested in learning about genetic services reporting they had noticed an increased impact. There was no significant difference found between responses for these measures and gender, age, practice setting or practice location.

Utilization of genetic services

The majority of respondents reported never or rarely referring patients to genetic counselors (72%, 113/157) or specialists other than a genetic counselor for genetic counseling and/or testing (68%, 106/157). Low percentages of participants reported referring to genetic counselors or non-genetic counselor specialists somewhat or very frequently (12%, 19/157 and 17%, 27/157, respectively). Ninety-one percent (143/157) reported never referring to telemedicine services. A much higher percentage of those who had not discussed genetics more in the last 5-10 years reported they never refer to a genetic counselor (63%, 27/43) than those who had discussed genetics more (19%, 15/79, p=0.001). Responses varied significantly by gender, with approximately twice the percentage of females reporting they refer to specialists for genetic counseling and testing somewhat or very frequently than males (24%, 15/62 versus 13%, 12/95, p=0.041).

Frequency of referral to genetic counselors varied significantly by practice setting (p=0.039), practice location (p=0.014) and specialty (p<0.001). Twenty-three percent (12/53) of participants in a university medical center setting reported referring to genetic counselors somewhat or very frequently, versus less than 10% of those in all other settings. Sixteen percent (14/87) of participants in an urban setting reported referring to genetic counselors somewhat or very frequently, versus 6% (3/49) of suburban and 5% (1/19) of rural participants.

There was a moderate correlation between the perceived integrality of genetics to patient care in the participants' specialty and frequency of referral to genetic counselors (Spearman's rho=0.51, p<0.001). Of those that felt that genetics was not at all integral, nearly all (94%, 34/36) never or rarely referred with the remainder occasionally referring. In contrast, among those who felt that genetics was

moderately or very integral, less than half never or rarely referred (45%, 30/66), with the rest split between occasionally (26%, 17/66) or somewhat/frequently (29%, 19/66) referring (Figure 2).

Participants were asked to select types of specialists they refer to for genetic counseling and/or testing. Forty-two percent (66/157) selected genetic counselor, 17% (27/157) selected maternal-fetal medicine, 35% (55/157) selected MD geneticist, 10% (15/157) selected high-risk oncologist, 3% (5/157) selected genetics nurse and 3% (4/157) selected other. Thirty-one percent (49/157) responded that they do not refer patients to other specialists for genetic testing.

Perceived Barriers to genetic services

Participants were given a list of potential barriers to referring a patient to a genetic counselor and asked to select all choices that applied. Table III delineates the frequency of responses chosen. The most commonly selected barrier (29%, 45/157) was that the participant referred to a physician specialist for genetics-related indications. Over one quarter (26%, 40/157) indicated genetic counseling was not indicated for their patients. About one-fifth of the respondents indicated awareness-related barriers such as not knowing any genetic counselors available to their patients (21%, 33/157), not knowing when a genetic counselor would be appropriate (21%, 32/157) and not knowing how to refer to a genetic counselor (19%, 30/157). Characteristics of participants who chose certain barriers are described below.

"Genetic counseling in not indicated for my patients"

Participants who chose this barrier were less likely to report they were very aware of genetic services in the area (10%, 4/40) than those who did not choose this barrier (26%, 30/117). Only 5% of those who chose this barrier were Ob/Gyns, versus 21% (24/117) of those who did not (p=0.019).

"I don't know any genetic counselors available to my patients"

Participants who selected this barrier were more likely to be interested in learning about genetic services available to their patients (97%, 32/33) than participants who did not choose this barrier (73%, 91/124, p=0.004). Only 6% (2/33) reported being very familiar with genetic counselor roles versus 33% (41/124) of those who did not choose this barrier (p=0.006). Participants who selected this barrier were also more likely to report they were not aware or were slightly aware of genetic services in their area (85%, 28/33) than those who did not select this barrier (44%, 54/124, p<0.001).

Table III Proportion of all participants selecting barriers

Barrier	% (n)
I refer to an MD geneticist or other physician specialists for genetics- related indications	29 (45)
Genetic counseling is not indicated for my patients	25 (40)
I don't know any genetic counselors available to my patients	21 (33)
I don't know when a genetic counselor would be appropriate	20 (32)
I don't know how to refer to a genetic counselor	19 (30)
My patients probably would not be able to afford it	18 (28)
I can order the appropriate genetic testing for my patients	17 (26)
I can perform the appropriate genetic counseling to my patients myself	10 (16)
There are no genetic counselors in my area	10 (15)
Other	9 (14)
The wait time for an appointment with a genetic counselor is too long	2 (3)
Genetic counselors are not MDs	1 (2)
I would have to refer to another doctor's office to reach a genetic counselor and don't want to risk losing my patient	<1 (1)

"I don't know when a genetic counselor would be appropriate"

Fewer participants who chose this barrier reported noticing an increase in the impact of genetics on their field in the last 5-10 years (63%, 20/32) than those who did not choose this barrier (76%, 95/125), but a higher percentage were interested in learning more genetic services available to their patients (94%, 30/32 versus 74%, 93/125). They were less likely to be familiar with the roles a genetic counselor can provide, with 63% (20/32) reporting they were not familiar or were slightly familiar versus only 34% (43/125) of those who did not choose this barrier reporting the same (p<0.001). They were also less likely to be aware of genetic services in the area, with 85% (27/32) reporting they were not aware or

were slightly aware in comparison with 44% (55/125) of those who did not choose this barrier reporting the same ($p<0.001$).

"I don't know how to refer to a genetic counselor"

Ninety-seven percent (29/30) of participants who indicated this barrier were interested in learning more about genetic services available to their patients, which was a higher percentage than the participants who did not indicate this barrier (74%, 94/127, $p=0.009$). They were less likely to report being very familiar with genetic counselor roles (3%, 1/30) than participants who did not choose this barrier (33%, 42/127, $p=0.002$). Lastly, more of these participants were not aware or were slightly aware of genetic services (87%, 26/30) and genetic testing (87%, 26/30) than participants who did not choose this barrier (44%, 56/127, $p<0.001$ and 40%, 51/127, $p<0.001$, respectively).

"I can perform the appropriate genetic counseling to my patients myself"

Participants who selected this barrier were more likely to be interested in learning about genetics and genetic testing as relevant to their field (94%, 15/16), more likely to know of a specific lab that performs genetic testing (88%, 14/16, $p=0.002$) and more likely to report being very familiar with the roles a genetic counselor can provide (75%, 12/16, $p<0.001$) than those who did not select this barrier. They were also more likely to be very aware of genetic services in the area (44%, 7/16, $p=0.009$), with none of these participants reporting they were not aware of services. The most common source of information for these respondents was electronic primary literature (75%, 12/16). Thirty eight percent (6/16) reported genetic counselors, a genetics laboratory/testing company and online websites as sources of information. Participants choosing this barrier were more likely to report spending at least 10 minutes providing genetic counseling (75%, 12/16) than those who did not choose this barrier (24%, 34/141, $p<0.001$).

"I can order the appropriate genetic testing for my patients"

Participants who selected this barrier were more likely to report that they were very familiar with genetic counselor roles (65%, 17/26, $p<0.001$) than those who did not choose this barrier (20%, 26/131). They were significantly more likely to be very aware of services in their area (42%, 11/26, $p=0.021$), very aware of genetic testing available to their patients (62%, 16/26, $p<0.001$) and know of a specific lab

that does genetic testing (88%, 23/26, $p<0.001$), than those who did not choose this barrier (18%, 23/131; 18%, 24/131; and 41%, 54/131; respectively). Participants who chose this barrier were more likely to report spending at least 10 minutes providing genetic counseling (65%, 17/26) than those who did not (18%, 29/160, $p<0.001$).

Discussion

Physicians surveyed in this study clearly acknowledge the increased impact of genetics in medicine, however, we have identified a substantial need for physician education regarding the roles of a genetic counselor, available genetic services and referral logistics. Furthermore, the majority of physicians surveyed reported the desire for education about genetics and genetic testing as relevant to their field. Specific results of this study can help direct efforts to educate non-genetics professionals to ensure more patients receive appropriate genetic evaluation and counseling, when indicated.

Large proportions of the participants overall reported no familiarity or only slight familiarity with genetic counselor roles and no awareness or only slight awareness of genetic testing and services, clearly demonstrating a need to educate physicians about genetic counselors and other genetic services available to their patients. Highlighting this need, publically available information revealed genetic counselors practicing in locations within ten miles of the zip codes of one-third of the participants who reported no genetic counselors in their area as a barrier to referral to genetic counselors. Genetic counselors were located within fifty miles of the zip codes of two-thirds of these participants. Therefore, physicians may perceive a lack of geographically accessible genetic counselors despite the fact that one may practice within fifty miles of their zip code.

More than half of participants did report they were moderately or very familiar with genetic counselor roles, but this study did not assess the accuracy of these perceptions. It is probable that many physicians have erroneous or incomplete perceptions of services provided by genetic counselors given previous literature demonstrating physicians often lack familiarity with complex genetic counseling issues such as ethical, legal and psychosocial implications (Demmer et al., 2000; Bensend et al., 2014). Physicians with a better understanding of the scope of practice of genetic counselors may be better equipped to identify patients that could benefit from meeting with a genetic counselor, therefore education about genetic counselors has great importance. A very high percentage of participants were not aware of telemedicine services. Although telemedicine services are sparse for a number of genetics issues, it will be essential to advertise these options as they become available since provision of genetic

services to patients in parts of the state far from major medical centers may depend solely on telemedicine technology.

A small group of participants indicated they do not refer to genetic counselors because they can perform the appropriate genetic counseling and order the appropriate testing themselves. Recalling previous research demonstrating physicians' general inability to perform these tasks raises the concern that patients may receive incomplete or incorrect information and services from physicians who counsel and order testing themselves (Brierley et al., 2010; Klitzman et al., 2013; Miller et al., 2014). Participants in this subset did indicate high familiarity with genetic counselors and reported spending more time providing genetic counseling to their patients than other participants. However, only about a third of these participants reporting spending 30 minutes or more providing genetic counseling in contrast with genetic counselors, who most often spend 45-60 minutes face-to-face with the patient (National Society of Genetic Counselors, 2014). Referring patients to genetic counselors would not only save physicians a significant amount of time but also afford patients more thorough and individualized counseling.

Furthermore, only half of all participants reported at least moderate awareness of genetic testing, but a high percentage of participants overall never or rarely refer to genetic counselors or other specialists for genetic testing. Therefore, more physicians may be ordering genetic testing without consultation with a genetics provider than the 17% who overtly reported doing so. Previous research demonstrates these physicians are at risk of failing to order appropriate tests, misinterpreting results and wasting healthcare dollars, emphasizing the need to increase awareness about the importance of involving a genetic specialist in the genetic testing process (Brierley et al., 2010; Miller et al., 2014).

About half of all participants also reported at least moderate awareness of genetic services. Considering again that the majority of participants never or rarely refer to genetic specialists, there exists discrepancy between reported awareness and utilization of services that, consistent with previous research, points to risk assessment as a missing element. If physicians lack the knowledge of genetics required to identify risk factors and make a referral, they may be unprepared to utilize genetics services effectively, even if they are aware of them (Baldwin et al., 2014; Leandro et al., 2014). This supports

basic genetics knowledge and risk assessment skills as important educational objectives. Increasing awareness of genetic services certainly remains essential, as half of all participants reported they were not aware or only slightly aware of services.

High percentages of participants across almost all specialties have noticed an increase in the impact of genetics on their field in the last 5-10 years, confirming the pervasiveness of the explosion of genetic discoveries in the last decade. However, fewer physicians reported genetics was integral to patient care in their specialty and fewer physicians reported discussing genetics more in their day-to-day practice. As Figure 1 illustrates, differences exist according to physician specialty. Participants with specialties in which genetics has traditionally played a larger role, Ob/Gyn and pediatrics, perceived genetics as more integral to their patient care than other specialties and also report discussing genetics more in their day-to-day practice. Interestingly, although relatively low percentages of family and internal medicine physicians feel that genetics is integral to patient care, they join Ob/Gyns and pediatricians in discussing it more day-to-day. Conceivably, the increase in discussion may have resulted from increased pressure or encouragement by medical associations for primary care physicians to ask certain family history screening questions (Heidelbaugh, & Tortorello, 2012; Qaseem et al., 2012). As shown by previous literature, however, physicians may not properly collect or assess family history information, and discussions regarding genetics may suffer from inaccurate and/or incomplete information (Baldwin, et al., 2014; Wood, Stockdale, & Flynn, 2008). Other specialties have also noticed an increased impact of genetics, but as a whole, they do not feel it is integral to patient care and are not discussing it more day-to-day. Perhaps physicians in these specialties have noticed that genetics discoveries have influenced the understanding of certain findings in their field but are not aware of potential benefits to patient care or do not know how to integrate genetics into their practice.

These specialty-dependent differences highlight that each group may warrant unique educational goals. Those who have noticed a recent increased impact and feel that genetics is integral to patient care in their practice would benefit from information about genetic services available to their patients and referral logistics, whereas those who have noticed an impact but do not feel genetics is integral to their

practice would benefit more from learning about identifying genetics indications, proper risk assessment and benefits of a genetic evaluation for the clinical care of certain patients.

Despite the majority of participants who had noticed an increased impact of genetics, the overall proportion who had not noticed an increased impact was surprisingly large, even when considering specialty differences. This alone indicates many physicians may be unaware of important genetic developments that could influence how they care for patients. For example, the dermatologists in the study who had not noticed an increased impact may be unaware that certain dermatologic findings are associated with genetic conditions for which evaluation and testing may be appropriate. A higher proportion of older participants answered they had not noticed an increased impact than younger participants, suggesting older physicians may be at greater risk of overlooking new information about genetics as related to their field.

A minority of participants overall felt genetics was integral to patient care in their practice, suggesting that even if physicians have noticed an increased impact of genetics on their field in some way, they often do not feel it plays a large role in how they currently care for patients. For many specialties represented in this study, genetics may not change how the physician directly cares for the patient, but it may be integral in guiding and improving the overall health care of the patient. In this case, the physician may have a responsibility to notice risk factors for genetic conditions and make the proper referrals. For example, an ophthalmologist who diagnoses a retinal achromic patch of a pediatric patient would disservice the patient by not recognizing the need for a genetic evaluation for tuberous sclerosis complex, which could identify potentially life-threatening health risks. As genetic contributions to common diseases are increasingly understood, primary care physicians may play an increasingly large role in genetic risk assessment and genetic testing for susceptibility to certain common diseases. They may also field results of population-based genetic screening that may potentially become implemented. Thus, it is important that physicians begin to appreciate the effect that genetics has and will have on patient care. This may start with basic genetics and genetic testing training and proceed to education regarding genetic services.

Figure 2 highlights that perceived integrality of genetics may drive participants' awareness and utilization of genetic services regardless of the participants' specialty. The trends suggest that the more integral a physician perceives genetics to be to patient care in his/her specialty, the more likely it is that he/she has noticed an increased impact, refers more frequently to genetic counselors and has greater awareness of genetic testing , genetic services and genetic counselor roles. Consider a subset of ten participants who designated they practice dermatology, a specialty with historically minimal genetics involvement. Four of these dermatologists felt that genetics was not at all or only slightly integral, had not noticed an increased impact and were not aware or only slightly aware of genetic services in their area. On the other hand, six of these dermatologists felt that genetics was moderately or very integral, had noticed an increased impact and were moderately or very aware of genetic services in their area. This insinuates that physicians who are less receptive to genetics issues in the first place may be less likely to notice an increased impact, even if it exists, or to be aware of genetic services available to their patients. This observation supports the need to determine a method to target physicians based on their perceived integrality of genetics to patient care in order to provide proper education.

In summary, this study reaffirms the need for education regarding basic genetics concepts, emphasizes the need for education regarding genetic counselors and services and identifies specific areas of focus for educational goals and outreach. Demographic characteristics did not significantly influence responses for most measures, signifying that physicians of all ages, gender, number of years practiced, practice setting and practice location could benefit from genetics education. The majority of participants' interest in learning more about genetics, genetic testing and genetic services is evidence that physicians are noticing an impact and desire engagement in the conversation regarding genetics issues.

Study Limitations

This study has several limitations worth noting. First, the study population generally reflected the gender and age breakdown of all Texas physicians but did not reflect the breakdown of specialties. Data from additional specialties, such as oncology, was not obtained and can be the focus of future research.

The survey employed was not a validated tool. Caution should be taken when drawing conclusions from the data. Much of this hinges on the potential ambiguity of questions and answer choices. For example, when answering the question, "Have you noticed an increase in the impact of genetics on your field in the last 5-10 years?" some participants may have only considered developments that have had a clinical impact, such as new clinically available genetic tests, whereas others may have considered aspects with varying degrees of clinical impact, such as exclusive drug trials for patients with certain genetic mutations or molecular genetic discoveries that provide explanations for findings relevant to their field but lack clinical significance at this time.

Research Recommendations

Future research could focus on surveying physicians in other specialties, especially oncology. Evaluating the nature of conversations physicians currently have with patients regarding genetics-related issues would identify potential strengths and areas for improvement. As genetic testing and screening become more widely available and utilized, whether through clinical offices, state-implemented programs or direct-to-consumer products, more research will be needed regarding the extent to which physicians are able to effectively handle genetics issues.

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