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Attitudes and Practices Among Internists Concerning Genetic Testing

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Abstract

Many questions remain concerning whether, when, and how physicians order genetic tests, and what factors are involved in their decisions. We surveyed 220 internists from two academic medical centers about their utilization of genetic testing. Rates of genetic utilizations varied widely by disease. Respondents were most likely to have ordered tests for Factor V Leiden (16.8%), followed by Breast/Ovarian Cancer (15.0%). In the past 6 months, 65% had counseled patients on genetic issues, 44% had ordered genetic tests, 38.5% had referred patients to a genetic counselor or geneticist, and 27.5% had received ads from commercial labs for genetic testing. Only 4.5% had tried to hide or disguise genetic information, and <2% have had patients report genetic discrimination. Only 53.4% knew of a geneticist/genetic counselor to whom to refer patients. Most rated their knowledge as very/somewhat poor concerning genetics (73.7%) and guidelines for genetic testing (87.1%). Most felt needs for more training on when to order tests (79%), and how to counsel patients (82%), interpret results (77.3%), and maintain privacy (80.6%). Physicians were more likely to have ordered a genetic test if patients inquired about

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genetic testing (p<.001), and if physicians had a geneticist/genetic counselor to whom to refer patients (p<.002), had referred patients to a geneticist/genetic counselor in the past 6 months, had more comfort counseling patients about testing (p<.019), counseled patients about genetics, larger practices (p<.032), fewer African-American patients (p<.027), and patients who had reported genetic discrimination (p<.044). In a multiple logistic regression, ordering a genetic test was associated with patients inquiring about testing, having referred patients to a geneticist/genetic counselor and knowing how to order tests., These data suggest that physicians recognize their knowledge deficits, and are interested in training. These findings have important implications for future medical practice, research, and education.

Keywords

genetic testing; medical education; doctor-patient communication; ethics; genetic discrimination; decision-making; genetic counseling

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Genetic information can potentially enhance the prevention and treatment of numerous disorders, but many questions exist concerning how physicians who are not geneticists view and utilize these tests – whether, when, how, and why they order tests. Genetic testing can potentially assist with diagnosis, guide treatment, and inform reproductive decisions. Genetic testing is available for genes associated with a growing number of disorders, from monogenic, highly penetrant conditions to monogenic conditions with low penetrance, and for susceptibility testing for several more common complex disorders. Yet especially for this last group of disorders, interpretations of genetic tests with relatively small increments in absolute risk require sophisticated knowledge that many primary care providers may lack. In the future, the number of genetic tests available and ordered by physicians will undoubtedly increase. Tests are being developed for a wide variety of disorders including cancer susceptibility, cardiac arrhythmias, seizures, immunodeficiencies, dementias, and pharmacogenetics. With news coverage of advances in genetics, and direct-to-consumer (DTC) marketing of tests, patients' interest in testing will no doubt continue to grow.

Recently, the director of the National Human Genome Research Institute called for enhancing physician use of genetic tests (Feero and Green 2011). Indeed, physicians can strongly affect patients' decisions about genetic testing, and play critical roles as gatekeepers to genetic assays (Klitzman et al. 2007; Klitzman 2009). Patients may also expect providers to have knowledge of and make appropriate referrals for genetic testing (Miller et al. 2010).

But only a limited number of studies have assessed physicians' uses of genetic testing, and this research suggests several problems. Physician knowledge of genetics has been low in self-reported surveys (Chase et al. 2002; Menasha et al. 2000) and in direct assessment (Finn et al. 2005). Only 37% regularly read articles on genetic testing (Menasha et al. 2000). For genetic testing for cancer susceptibility, physicians have indicated needs for clinical guidelines (89%), with many expressing concerns about insurance discrimination (81%) and confidentiality (53%) (Freedman et al. 2003). Most (95%) believe that physician responsibilities include counseling patients about genetic testing, but only 51% have time to do so (Escher and Sappino 2000; Menasha et al. 2000), and among psychiatrists, fewer than 25% felt prepared to engage in counseling (Finn et al. 2005).

Nonetheless, of US primary care physicians (PCPs), one study has suggested that 60% have ordered genetic tests, and 74% have referred a patient for genetic testing (Shields et al. 2008). Several factors may be involved in whether physicians order tests, including patient inquiry about testing, provider assessment of the probability of a patient's carrying a mutation, and practice environment, with participation in an integrated health system

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associated with increased use of testing (Sifri et al. 2003). Referral for cancer susceptibility tests was associated with patient request and physicians receiving genetic test advertising (Wideroff et al. 2003).

Physicians may under- or over-utilize genetic tests. Of 171 physicians who had treated Alzheimer's disease, knowledge about the proper use of *APOE* and *PS1* testing for both predictive and diagnostic purposes was poor, with more than half of the physicians in many scenarios endorsing genetic testing without appropriate justification (Chase et al. 2002). Primary care physicians may also order BRCA testing on patients who are at low risk and thus not appropriate for such referral (Bellcross et al. 2011; Trivers et al. 2011; Van Riel et al. 2010; White et al. 2008). Concerning hypothetical use of genetic tests for diabetes, 79% of PCPs and endocrinologists would be likely to order the tests, with 39% somewhat/very likely to do so prior to "published evidence of clinical efficacy" (Grant et al. 2009). Robust data to establish clinical utility may take several years to acquire and disseminate, and physicians may order tests before clinical utility has been established. The possibility of pharmacogenomics has also received attention, and 98% of physicians believed genetic profiles may eventually impact effectiveness of pharmaceutical therapy (Stanek et al. 2009). Yet pharmacogenomic tests for medications such as warfarin, tamoxifen, and anti-depressants have not been widely adopted by physicians.

Questions remain about what factors (e.g., related to characteristics of physicians or their patients) may be associated with physicians' practices concerning genetic testing for patients. Patient sociodemographics may play a role. Physicians serving more than 50% minority patients were significantly less likely to have ever ordered genetic tests for 3 of 4 conditions examined (breast/ovarian cancer, colon cancer, or Huntington's disease, but not sickle cell), or to refer patients for genetic testing (Shields et al. 2008).

Concerns about possible genetic discrimination may also potentially affect physicians' decisions about genetic tests, but how often physicians have observed or tried to prevent genetic discrimination is unknown. Some critics have argued that concerns about discrimination are exaggerated (Nowlan 2002; 2003; Wertz 2002). Hall and Rich (2000) suggested that presymptomatic individuals experience little or no discrimination in obtaining health insurance, but these researchers interviewed mostly individuals who worked in the insurance industry. In a 2004–2006 a study of California physician and nurses, most believed that fears of discrimination would deter patients from genetic testing, but most respondents were unaware of state and federal laws prohibiting health insurance discrimination (Lowstuter et al. 2008).

The US Genetic Information Nondiscrimination Act (GINA) was enacted in 2008, yet key questions remain about how physicians and patients since then view the possibility of genetic discrimination, and what, if anything, they may do as a result (e.g., whether their concerns affect their willingness to order genetic testing, and/or efforts to disguise genetic information). Many of the prior studies above were done over a decade ago. The amount of genetic research and testing has mushroomed since then, and almost all of these studies were conducted prior to the passage of GINA.

Given that rapid technological advances will offer more extensive testing and reduce its cost, understanding internists' and PCPs' knowledge, attitudes, and practices regarding genetic testing and factors involved is crucial. Clinical guidelines for utilization of genetic testing are increasingly being developed (EGAPP Working Group 2011; Fatkin et al. 2007; Goldman et al. 2011; Gollob et al. 2011; Hershberger et al. 2009), but it is unclear how many physicians are aware of these guidelines, or in what specific areas they see themselves as needing training.

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The ever-rising number of available tests, identifying genetic markers with very different degrees of predictiveness and clinical utility, may exacerbate these problems. Yet prior studies have each focused on testing for only one or a few disorders – particularly *BRCA 1/2*, with some attention to Huntington's disease, hereditary nonpolyposis colorectal cancer, and a few other diseases (Freedman et al. 2003; Plon et al. 2011; Prochniak et al. 2011; Shields et al. 2008; Sifri et al. 2003; Trivers et al. 2011; Van Riel et al. 2010), or they have not specified any diseases (Haga et al. 2011). Yet over 2,200 genetic tests exist. Hence, critical questions also emerge as to whether physicians differentiate between different tests in ordering practices, and if so, to what degree and how.

Thus, the purpose of this study was to explore these realms – specifically whether internists order genetic tests on their patients, and if so, how often, and for which diseases, and what factors (e.g., characteristics of the provider, and his/her patients) may be associated with these decisions.

Methodology

Sample and Procedures

We recruited internists who were members of departments of Internal Medicine at two academic medical centers. Invitations to participate in the survey were sent by email with a link to the survey instruments in July 2009 and two reminders were sent to all department members by email. We offered a $5 gift certificate at a national coffee store chain. The email lists contained approximately 993 and 110 addresses, respectively. Of 1103 names on the combined lists, 220 physicians responded, yielding a conservative estimated 19.9% response rate.

Instrumentation

The survey instrument was a 44-item questionnaire, developed by the investigators based on past published literature and clinical experience, accessible through the online survey system Survey Monkey (www.surveymonkey.com). We examined several domains: 1) clinicians' self-reported knowledge of genetics and genetic testing in their area of practice; 2) clinicians' views and practices concerning genetic testing and genetic privacy; 3) barriers and facilitators to appropriate clinician use of genetic tests, including clinician characteristics (e.g., profession, specialty, number of years since completion of training, type of practice, gender, ethnicity, religion), and patient population characteristics as reported by these physicians (e.g., ethnicity, type of insurance coverage); and 4) self-identified needs and implications for continuing education, clinical guidelines, and further assistance in this area. We pilot tested the survey with five clinicians first and revised accordingly prior to distribution.

Data Analysis

Results from the survey were transferred from the Survey Monkey platform to a database for use in SPSS. Statistical analyses included chi-square tests for examination of categorical variables. We also conducted a multiple logistic regression to explore which independent variables were most associated with whether physicians had ordered a genetic test or not. We simultaneously entered into the model variables that were significantly associated with the outcomes in Chi square tests (as reported on Table 3). As seen on Tables 1 and 3, some participants did not answer all questions (e.g., only 77% answered whether they had personally had a genetic test done, or whether patients had ever reported genetic discrimination). Hence, calculations are based on those participants who responded to each question.

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Results

As seen in Tables 1, 2, and 3, the sample was diverse in sex, age, ethnicity, specialties, work settings of providers, and insurance of the patient population. Rates of ordering tests varied widely by disease. Respondents were most likely to have ordered tests for Factor V Leiden (16.8% n=36), followed by breast/ovarian cancer (15.0%, n=32), and hypertrophic cardiomyopathy (8.4%, n=18). Overall, in the past 6 months, most (65%, n=115) reported having counseled patients on genetic issues, and many respondents had ordered genetic tests (44%, n=77), referred patients to a genetic counselor or geneticist (38.5%, n=67), and received ads from commercial labs for genetic testing (27.5%, n=49). Only half indicated they had "a geneticist or genetic counselor to whom you can refer patients" (53.4%, n=93), 4.5% (n=8) had taken steps to hide or disguise genetic information, and few (1.8%, n=3) have had patients report genetic discrimination.

Over half of the respondents thought that genetic testing should be performed on more patients in their specialty (54.4%), and that they should spend more time talking with their patients about family histories (57.3%), but 73.7% rated their knowledge of genetics as very/somewhat poor, and 87.1% rated their knowledge of guidelines for genetic testing as very/somewhat poor. Most indicated a need for more training on when to order genetic testing (79%), and how to counsel patients (82%), interpret results of genetic tests (77.3%), and maintain genetic privacy (80.6%). Physicians favored additional genetic education on-line (90%), in preference to grand rounds (86.3%), through the mail (i.e., printed materials sent via the US Postal Service) (64.4%), and/or at day-long conferences (45%).

Chi-Square Analyses

As shown on Table 3, physicians were more likely to have ordered a genetic test if patients had inquired about genetic testing (62.5% v. 24.4%, p<.001), and if physicians had larger practices (66.7% v. 29.7%, p<.032), had fewer African-American patients (55.8% v. 37.3%, p<.027), were comfortable counseling patients about genetic testing (62.5% v. 39.7%. p<.019), had a geneticist or geneticist/genetic counselor to whom to refer patients (53.8% v. 30.9%, p<.002), had patients who reported genetic discrimination (100.0% v. 41.9%, p<.044), and had knowledge of how to order and where to send tests (p<.029). There were trends for those who ordered genetic tests to be male (51.2%, 37.6%, p<.071), have more Caucasian patients in their practice (56.1% v. 39.0%, p<.056), and have received ads from genetic testing laboratories (55.1% v. 39.5%, p<.062). Physicians' decade of completion of medical training, ethnicity, having had a genetic test, patients, concerns about genetic privacy, having tried to hide/disguise genetic information, and perceived needs for training, and patients' types of insurance were all not significantly associated with ordering a genetic test.

Binary Logistic Regression Analysis

We individually entered into a simple binary logistic regression model all of the variables that we found in univariate analyses, as indicated above, to be significantly associated with having ordered a genetic test. All variables found to be significant in the simple binary logistic regression model were then simultaneously entered into a multiple binary logistic regression model. As seen on Table 4, the multiple binary logistic regression indicated that ordering a genetic test was associated with: patients asking about testing (p<.021; OR, 3.22; 95% CI 1.190–8.709), having referred patients to a geneticist/genetic counselor in the past 6 months (p<.01; OR, 4.5; 95% CI, 1.590–12.739), and having counseled patients on genetic issues in the past 6 months (p<.01 OR, 4.62; 95% CI, 1.422–15.031).

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Discussion

These data, the first to examine several critical areas, including rates of internists ordering several specific genetic tests, disguising genetic information, and having patients report discrimination, highlight several key issues. These data reveal considerable continuing self-reported deficits in knowledge of genetic testing and guidelines – even among highly-trained physicians in respected academic medical institutions, with innumerable opportunities for continuing medical education. Specifically, the fact that the substantial majority (73.7% and 87.1% respectively) rated their knowledge of genetics and of guidelines for testing as very/somewhat poor poses concerns. Moreover, large numbers who rated their knowledge of genetics and of guidelines as very/somewhat poor (41.1% and 42.3% respectively), and were very/somewhat uncomfortable ordering/referring for testing and counseling patients (26.1% and 39.7% respectively), still ordered genetic tests. The fact that physicians may be ordering tests without sufficient knowledge or comfort suggests that these tests may be ordered in suboptimal ways, highlighting crucial needs for more targeted education. These results thus underscore needs to substantially enhance provider understandings of genetics and guidelines, and comfort counseling patients. Future research is also critical to probe more fully the barriers that exist in each of these areas.

On the positive side, these physicians widely endorsed the need for more training in all four areas probed: how to order genetic tests, counsel patients, interpret test results, and maintain genetic privacy. Thus, amid calls for increased physician use of genetic assays (Feero and Green 2011), the present data highlight the breadth of tests now used by at least some physicians, but also the challenges these providers face in genetic test utilization and interpretation.

We examined a far wider array of diseases than have prior studies of physicians. Importantly, while prior studies have focused heavily on cancer susceptibility (particularly BRCA and colorectal cancer [Freedman et al. 2003; Plon et al. 2011; Shields et al. 2008; Sifri et al. 2003)], HD, and sickle cell (Shields et al. 2008), we examined several other conditions as well and found that the most commonly ordered test was none of these, but Factor V Leiden, which is often part of a coagulopathy evaluation. In the present study several other tests were commonly ordered as well (e.g., hypertrophic cardiomyopathy, cystic fibrosis, hemochromatosis, and pharmacogenetic tests), though specific physician uses of these test results have not been examined. These data suggest internists tend to test for a variety of treatable diseases, and that future studies should investigate a far broader range of tests than heretofore.

The wide differences in experiences ordering genetic tests are in some ways reassuring, given differences in disease severity, preventability, treatability, age at presentation, and purpose of testing (diagnosis, susceptibility, or carrier status), but they suggest several challenges for future practice and research. Efforts to understand providers' decisions regarding genetic assays need to account for variations in test accuracy and reliability, and in the purpose of testing. These findings also highlight difficulties in making generalizations about physicians' attitudes and practices concerning genetic testing, given the wide variation in the purpose and clinical utility of different tests. Clearly, genetic tests do not constitute one monolithic category, but vary widely in indications (diagnostic confirmation, predictive testing, carrier screening, and pharmacogenetic tests) and use.

The logistic regression showed that factors related to patient behavior, and physician knowledge and behavior were most strongly associated with ordering a genetic test. Having fewer African-American patients was not significant in the regression, possibly because it may have been reflected in patients inquiring about tests (i.e., patients may vary in whether they inquire about testing based on several factors, including their ethnicity). Patient report

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of genetic discrimination was also not significant in the regression, probably since few physicians overall heard such reports. Physician comfort with genetic counseling may have been reflected in physicians knowing how to order/where to send tests. Having a larger practice was probably subsumed under having had patients ask about testing.

The fact that in the present study, ordering tests was associated with patient inquiry and patients' ethnicity raises concerns about the rational use of these assays. Ordering a genetic test in the preceding six months was most strongly associated with patient inquiry about genetic testing, as Sifri et al. (2003) found as well. Yet this finding is potentially troubling, in part given the growth of marketing of genetic tests directly to consumers, even for specific tests (Myers and Jorgensen 2005). The fact that the likelihood of a physician ordering a genetic test may be driven in part by a patient asking about it suggests that physicians' decisions about ordering genetic tests may be driven by factors other than clinical indications or clinical utility. Granted, a patient may be interested in genetic results, even when these are not clinically actionable, simply to provide peace of mind or make informed life-planning decisions. But some patients may ask about a test, mistakenly thinking that they are at high risk for a disease, or believing that a negative genetic test will mean they are not at risk at all. Hopefully, most physicians order tests as a result of collaborative, informed decisions rather than simply to placate uninformed or misinformed patients.

The fact that only half of physicians had a geneticist or genetic counselor to whom they could refer patients also raises concerns. The finding that ordering of tests was significantly associated with having such an expert available highlights the vital role that such referral sources can play. Limitations in insurance reimbursement no doubt hamper these referrals (i.e., genetic counseling is typically not covered by health insurance plans), underscoring needs for changes in policy to address these impediments to care.

While Shields et al. (2008) found that providers with >50% minority patients were less likely to order 3 of 4 tests examined, we found that providers with higher proportions of African-American patients, but not of Hispanic patients, were less likely to have ordered a genetic test. Hence, the specific type of minority patients a provider treats may affect rates of testing. This finding is of concern, however, as it suggests that certain groups may be offered potentially beneficial testing less than others, exacerbating existing health inequities. At the same time, some minority communities are significantly more concerned about disadvantages and potential abuses of genetic testing, which may lead to less interest and demand among certain groups (Forman and Hall 2009; Susswein et al. 2008; Thompson et al. 2003). We did not examine whether the rates of patients requesting genetic testing differed between different ethnicities, or whether minority patients were offered genetic testing and subsequently declined, but future research should do so.

Few physicians in our post-GINA survey have observed genetic discrimination or taken steps to disguise genetic information. Less than 2% of the internist respondents had heard reports of discrimination from their patients. Yet in 1996, 25% and 22% of members of a variety of genetic support groups reported that their family members were refused life insurance or health insurance, respectively (Lapham et al. 1996), and 10% of genetic clients in Australia have reported discrimination (mostly in life insurance – in part since Australia has national health insurance) (Taylor et al. 2007; Taylor et al. 2008). Clearly, different types of discrimination can exist (life insurance, employment, etc.), and may affect family members, rather than patients themselves. Internists may not inquire about these domains, and discrimination may be subtle (Klitzman 2010), such that physicians and even patients are not aware of it. Nonetheless, it is important that providers be aware of the possibility of discrimination, in order to document and address it, if and when it occurs.

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Only 4.5% of the physicians in this study have taken steps to disguise genetic information. Anecdotally, patient support groups for HD and other diseases have advocated that patients ask doctors not to record sensitive genetic information in the medical record for patients who are presymptomatic. Yet disguising test results, while reducing concerns about privacy, can potentially be detrimental to medical care. The low percentage of physicians who acknowledged disguising such information may be because this group of physicians is rarely ordering predictive genetic testing on asymptomatic patients for genetic conditions, such as HD, where potential discrimination may be most likely. Especially with increasing use of electronic medical records (EMRs), future studies should continue to assess such practices among physicians over time.

These data have several additional critical implications for future research and practice, for instance, suggesting needs to examine advertising for commercial genetic testing (what tests get advertised, to whom, and what these advertisements claim about the utility and potential limitations, if any, of genetic testing). Advertising could fill an educational gap, if it correctly informs providers about newly available genetic tests, clinical indications, and utility. The fact that 80% of physicians wanted more training about how to maintain genetic privacy is of importance, given the widening adoption of EMRs, which, by making information more easily available to treaters, may affect physician and patient views about potential discrimination. Efficient EMR systems could also potentially provide just-in-time education about genetic testing for relevant patients through pop-up windows, identifying patients who are candidates for genetic testing, based upon the information in the EMR.

Study Limitations

This study has several potential limitations. Compared to physicians as a whole nationally (Runy 2009), our sample of respondents did not differ in terms of ethnicity, but did include more women (53.3%, compared with 28.26% nationally). This difference may be due to our focus on internists, which may include high numbers of primary care physicians who may be more likely to be women. Indeed, since 1990, more women than men specialize in family practice (Lambert and Holmboe, 2005), and in 2005, 49.5% of primary care physicians were women (Tu and O'Malley, 2007). In surveying primary care physicians about BRCA testing, Escher et al. (2000) similarly had a significantly higher response rate from women than men (76% vs. 60% $p < .008$). Several other articles reporting on physician attitudes and practices concerning genetic testing did not examine gender of respondents in any way (Friedman et al. 2003; Grant et al. 2009; Lowstuter et al. 2008; Prochniak et al 2011; Shields et al. 2008). Female physicians may be more personally aware of and concerned about these issues, though in the present study gender was not significantly associated with whether respondents had ordered genetic tests or not. Future research is clearly needed to explore these relationships more fully

We included physicians only from academic internal medicine departments in two academic medical centers in a single geographic region, and the response rate was 19.9%. Yet this initial exploration of many of these issues still found several suggestive findings that can be further investigated in future research with larger and more diverse samples. The surveys also relied on self-reports. We did not investigate whether the tests they ordered were appropriate for each clinical situation. Given the relatively small sample size, we also did not explore other potential associations (e.g., whether certain groups of specialists were more likely to order specific tests). Owing to the exploratory nature of the present study, we did not adjust the p-values on the Chi-squared tests to account for multiple comparisons. However, future studies can probe these issues in further detail among larger samples.

In sum, these data have critical implications for medical practice, research, policy, and education in this rapidly expanding area. For example, they highlight several areas for future

studies and targeted educational efforts, and needs for policies that mandate wider reimbursement for genetic counseling.

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Table 1

Characteristics of Respondents

Characteristic	% (*n*)
Gender	
Male	46.7% (100)
Female	53.3% (114)
Age	
20–29	4.7% (10)
30–39	45.3% (97)
40–49	20.1% (43)
50–59	15.0% (32)
60+	15.0% (32)
Year of Med School Graduation	
Before 1990	36.4% (76)
After 1990	63.6% (133)
Specialty	
General Medicine	19.6% (42)
Cardiology	15.4% (33)
Pulmonary/Critical Care	9.8% (21)
Digestive & Liver Disease	9.8% (21)
Endocrinology	8.9% (19)
Hematology/Oncology	8.4% (18)
Other	7.5% (16)
Nephrology	7.0% (15)
Infectious Disease	3.7% (8)
Family Medicine	3.7% (8)
Emergency Medicine	3.3% (7)
Rheumatology	2.8% (6)
Workplace Setting	
Hospital-based	77.4% (164)
Community-based	8.5% (18)
Combination of above	14.2% (30)
MD Ethnicity	
White	68.5% (146)
Asian/Pacific Islander	21.1% (45)
Hispanic	4.2% (9)
Other	3.4% (8)
African-American	2.8% (6)

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Table 2

Genetic Tests Ordered in Past Six Months

Ordering Tests/Referring for Specific Diseases	***n*** **(%)**
Factor V Leiden	36 (16.8%)
Breast/Ovarian Cancer	32 (15.0%)
Hypertrophic Cardiomyopathy	18 (8.4%)
Hereditary Nonpolyposis Colorectal Cancer	17 (7.9%)
Sickle Cell	17 (7.9%)
Cystic Fibrosis	13 (6.1%)
Drug Toxicity/Pharmacogenetics	12 (5.6%)
Hereditary Hemochromatosis	12 (5.6%)
Chromosome Analysis	11 (5.1%)
Familial Adenomatous Polyposis	10 (4.7%)
Inherited Arrhythmias	10 (4.7%)
Methylenetetrahydrofolate Reductase	7 (3.3%)
Hearing Loss	4 (1.9%)
Alzheimer’s Disease	3 (1.4%)
Fragile X	2 (0.9%)
Psychiatric Illness	2 (0.9%)
Tay Sachs	2 (0.9%)
Epilepsy	1 (0.5%)
Huntington’s Disease	1 (0.5%)
Spinal Muscular Atrophy	1 (0.5%)

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Table 3

Characteristics of Internists Who Ordered a Genetic Test in Past 6 Months

	Ordered Genetic Test (N v. Y)		
	No % (*n*)	**Yes % (*n*)**	***p***
MD Demographics			
Gender			0.071
Male	48.8% (40)	51.2% (42)	
Female	62.4% (58)	37.6% (35)	
Age			0.084
20–29	62.5% (5)	37.5% (3)	
30–39	52.9% (45)	47.1% (40)	
40–49	74.2% (23)	25.8% (8)	
50–59	37.5% (9)	62.5% (15)	
60+	60.7% (17)	39.3% (11)	
Completed medical training decade			NS
1960–1969	80.0% (4)	20.0% (1)	
1970–1979	60.0% (9)	40.0% (6)	
1980–1989	40.0% (10)	60.0% (15)	
1990–1999	60.9% (14)	39.1% (9)	
2000–2009	59.1% (52)	40.9% (36)	
2010–2019	14.3% (1)	85.7% (6)	
Still in training	50.0% (1)	50.0% (1)	
Ethnicity			NS
White	54.9% (67)	45.1% (55)	
African-American	50.0% (3)	50.0% (3)	
Asian-Pacific Islander	56.4% (22)	43.6% (17)	
Hispanic	66.7% (4)	33.3% (2)	
Other	80.0% (4)	20.0% (1)	
Patient Demographics			
Percent White			0.056
<50%	61% (75)	39% (48)	
>50%	43.9% (18)	56.1% (23)	
Percent African-American			0.027
<10%	44.2% (23)	55.8% (29)	
>10%	62.7% (69)	37.3% (41)	
Percent Asian Pacific Islander			0.081
<5%	66% (33)	34% (17)	
>5%	50% (35)	50% (35)	
Percent Hispanic			NS
<25%	52.9% (27)	47.1% (24)	

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	Ordered Genetic Test (N v. Y)		
	No % (*n*)	**Yes % (*n*)**	***p***
>25%	58.3% (67)	41.7% (48)	
Work setting			0.096
Hospital-based	56.5% (78)	43.5% (60)	
Community-based	35.3% (6)	64.7% (11)	
Combination of hospital/community-based	69.6% (16)	30.4% (7)	
Number active patients in practice			0.032
Less than 100	70.3% (26)	29.7% (11)	
100–499	53.4% (31)	46.6% (27)	
500–1000	62.5% (20)	37.5% (12)	
Over 1000	33.3% (8)	66.7% (16)	
Percent of Medicaid patients			NS
<25%	52.2% (35)	47.8% (32)	
>25%	57.6% (53)	42.4% (39)	
Percent of Medicare patients			NS
<25%	53.4% (31)	46.6% (27)	
>25%	57.8% (59)	42.2% (43)	
Percent patients with private insurance			NS
<25%	63.6% (42)	36.4% (24)	
>25%	53.8% (43)	46.3% (37)	
Percent patients with no insurance			NS
<5%	52.3% (34)	47.7% (31)	
>5%	64.2% (43)	35.8% (24)	
MD/Patient Experience with Genetic Testing			
Received ads from labs			0.062
No	60.5% (78)	39.5% (51)	
Yes	44.9% (22)	55.1% (27)	
Pts asked about genetic testing in past 6 mo.			<0.001
No	75.6% (65)	24.4% (21)	
Yes	37.5% (33)	62.5% (55)	
How concerned about genetic privacy			NS
Not very concerned	61.0% (25)	39.0% (16)	
Somewhat concerned	56.2% (50)	43.8% (39)	
Very concerned	53.2% (25)	46.8% (22)	
Use method to hide/disguise genetic information			NS
No	57.1% (96)	42.9% (72)	
Yes	37.5% (3)	62.5% (5)	
Pts reported genetic discrimination			0.044
No	58.1% (97)	41.9% (70)	

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	Ordered Genetic Test (N v. Y)		
	No % (*n*)	**Yes % (*n*)**	***p***
Yes	0% (0)	100.0% (3)	
MD Experience with Genetic Testing/Counseling			
Have counseled pts on genetic issues past 6 mo.			<0.001
No	77.4% (48)	22.6% (14)	
Yes	45.2% (52)	54.8% (63)	
Have referred pts to geneticist/genetic counselor past 6 mo.			<0.001
No	72.0% (77)	28.0% (30)	
Yes	29.9% (20)	70.1% (47)	
Have geneticist/genetic counselor for referral			0.002
No	69.1% (56)	30.9% (25)	
Yes	46.2% (43)	53.8% (50)	
Current practice genetic testing performed			NS
More often	56.4% (53)	43.6% (41)	
Less often	50.0% (6)	50.0% (6)	
About the same	58.2% (39)	41.8% (28)	
Spend more time on pts family histories			NS
No	58.9% (43)	41.1% (30)	
Yes	53.5% (53)	46.5% (46)	
MD Knowledge of Genetic Testing			
Knowledge of genetics			NS
Very poor/Somewhat poor	58.9% (76)	41.1% (53)	
Somewhat good/Good/Very good	46.5% (20)	53.5% (23)	
Knowledge of guidelines pts genetic testing			NS
Very poor/Somewhat poor	57.7% (86)	42.3% (63)	
Somewhat good/Good/Very good	40.9% (9)	59.1% (13)	
Personally had genetic test			NS
No	54.7% (70)	45.3% (58)	
Yes	58.1% (25)	41.9% (18)	
Comfort ordering/referring testing to pts			0.073
Very uncomfortable/Somewhat uncomfortable	73.9% (34)	26.1% (12)	
Somewhat comfortable/Comfortable/Very Comfortable	53.6% (15)	46.4% (13)	
Comfort counseling pts on genetic testing			0.019
Very uncomfortable/Somewhat uncomfortable	60.3% (85)	39.7% (56)	
Somewhat comfortable/Comfortable/Very Comfortable	37.5% (12)	62.5% (20)	
MD Views on Genetic Testing Training			
More training when to order genetic testing			NS
No	51.4% (19)	48.6% (18)	
Yes	57.4% (78)	42.6% (58)	

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	Ordered Genetic Test (N v. Y)		
	No % (*n*)	**Yes % (*n*)**	***p***
More training how to order genetic testing			NS
No	46.7% (14)	53.3% (16)	
Yes	58.0% (83)	42.0% (60)	
More training counseling pts on genetic testing options			NS
No	56.3% (18)	43.8% (14)	
Yes	56.0% (79)	44.0% (62)	
More training interpreting test results			NS
No	53.8% (21)	46.2% (18)	
Yes	56.7% (76)	43.3% (58)	
More training maintaining genetic privacy			NS
No	51.4% (18)	48.6% (17)	
Yes	57.7% (79)	42.3% (58)	
Genetic education day-long conference			NS
No	59.6% (59)	40.4% (40)	
Yes	50.0% (34)	50.0% (34)	
Genetic education grand rounds			NS
No	63.0% (17)	37.0% (10)	
Yes	53.5% (76)	46.5% (66)	
Genetic education mailed information			NS
No	53.3% (32)	46.7% (28)	
Yes	56.5% (61)	43.5% (47)	
Genetic education online information			NS
No	37.5% (6)	62.5% (10)	
Yes	57.3% (86)	42.7% (64)	
Knowledge of how to order and where to send tests*			0.029
One a scale of 1–5 (with 1 the least and 5 the most)	3.15 ± 1.30	2.68 ± 1.45	

*Note: T-test used in this univariate analysis (Mean and standard deviation was reported for this continuous variable. The corresponding p-value indicated by T-test is for the comparison of the two groups). Chi-squared used in all other univariate analyses in this table.

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Table 4

Predictors of Having Ordered a Genetic Test in the Past 6 Months - Multiple Logistic Regression Analysis

Predictor	OR	95% CI	p value
Have referred patients to a geneticist or genetic counselor	4.501	1.590–12.739	< 0.01
Have counseled patients on genetic issues	4.623	1.422–15.031	< 0.01
Patients asked about genetic testing	3.22	1.190–8.709	= 0.02
Lack knowledge on how to order and where to send tests	0.741	.465–1.180	= 0.21
Insufficient knowledge or experience about genetic tests	1.418	.801–2.509	= 0.23
Lack of clinical guidelines	0.772	.506–1.178	= 0.23
Number of patients in practice	1.283	.811–2.030	= 0.29
Comfort counseling patients	1.857	.513–6.716	= 0.35
Have a geneticist or genetic counselor for referral	0.652	.220–1.927	= 0.44
Percent of African American patients	0.984	.354–2.735	= 0.98