

# Genetic Testing: A Physician's Perspective

JOSHUA D. MENASHA, M.D.<sup>1</sup>, CLYDE SCHECHTER, M.D.<sup>2</sup>, AND JUDITH WILLNER, M.D.<sup>3</sup>

## Abstract

Progress in DNA diagnostics has been extremely rapid. We sought to determine attitudes, awareness, and knowledge of genetic testing by physicians affiliated with the Mount Sinai Medical Center.

We surveyed 363 physicians within whose fields genetic testing for various diseases and disorders exist. Physicians' awareness of and opinions regarding testing, attitudes toward counseling, knowledge of the field, and interest in further education were assessed. Three hundred forty-one (341) physicians were determined to be eligible for the study and, of these, 89 (26%) returned completed surveys.

Of the respondents, 71% rated their knowledge of genetics and genetic testing as "fair" to "poor"; only 37% read articles concerning genetic testing on a regular basis. Physician awareness of currently available testing produced a bell-shaped distribution. Knowledge regarding Mendelian genetics yielded a bimodal distribution, and knowledge reflecting an understanding of the mechanics behind genetic testing produced a bell-like curve, skewed to the right. Those who identified themselves as practicing within an "academic" setting scored significantly higher on the Mendelian genetics and testing mechanics sections than those practicing in a "private" setting. Ninety-eight percent (98%) of the physicians said they would refer their patients to a genetic counselor. Although 91% of the respondents were aware of the existence of genetic counseling services, only 71% were aware of the services available at major New York medical centers. Of those aware of counseling services, 53% had referred a patient to them, and 83% of those who referred were "mostly" to "very" satisfied with the counseling. Ninety-five percent (95%) of the physicians believed that the doctor, among others, has the responsibility to counsel patients about genetic testing, yet only 51% felt that they had the time. No statistically significant preference was found concerning the methods for gaining further education or information about genetic testing.

Further education for physicians is required in order for them to accurately convey the risks and benefits of genetic testing to their patients. Furthermore, awareness of the counseling services available within the New York area needs to be heightened in order to provide physicians and patients with the specific services they desire. The most efficient and effective methods for providing information and for heightening awareness need to be determined through additional research. **Key Words:** Genetic testing, genetic counseling, physician knowledge.

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AFTER A DECADE OF RESEARCH, the international Human Genome Project is turning the idea of mapping the 80,000 to 100,000 genes which compose the human genome into a reality (1). With such information, geneticists will be able to identify, map, and sequence genes specific to or associated with numerous diseases, potentially leading to the development of genetic tests for these diseases (1). Testing has already become commercially available to physicians for a variety of can-

cers and single gene disorders (e.g., colon cancer and cystic fibrosis), and more tests are currently in development (2, 3). A proper understanding of the test results, however, is essential not only for making an accurate clinical diagnosis, but also for counseling the patient, who must clearly understand and prepare for the potential consequences of the results. The field of genetic testing has begun to change the practice of medicine substantially, as patients at risk for certain diseases can be identified well before clinical signs and symptoms manifest themselves. Furthermore, current definitions of "standard of care" and "duty" to one's patients and their families must be redefined in light of the new information this technology provides (4).

Only recently have studies been carried out to assess the opinions, awareness, and knowledge currently held by a variety of physicians regarding genetic testing (3, 5–8). Only with the data gained from such studies can physicians' strengths and shortcomings in this area be ascertained. Then, new information can be distributed to them in an optimal manner, to prepare them for

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<sup>1</sup>Resident, Departments of Human Genetics and Pediatrics, <sup>2</sup>Associate Professor, Department of Community and Preventive Medicine, and <sup>3</sup>Associate Professor, Departments of Human Genetics and Pediatrics, Mount Sinai School of Medicine, New York, NY.

From the Departments of Human Genetics and Community Medicine, Mount Sinai School of Medicine, New York, NY.

Address all correspondence to Joshua Menasha, M.D., Department of Pediatrics, Box 1512, Mount Sinai School of Medicine, One East 100th Street, New York, NY 10029.

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the constantly evolving and ever-growing emphasis on genetic technology in medicine. This paper hopes to present such data as it relates to the physicians who practice at the Mount Sinai Medical Center and its affiliates.

## Methods

### Subjects

Physicians were identified through lists obtained from their various departments. These lists represent physicians practicing at the time of the study, at either the Mount Sinai Hospital or an affiliate, when applicable. Departments were chosen if they dealt with diseases for which genetic testing was currently available. Three hundred sixty-three (363) physicians were initially identified from the departments of Neurology (n = 84, 23% of those surveyed), Pulmonary Medicine (n = 13, 4%), Geriatrics (n = 33, 9%), Cardiology (n = 36, 10%), Oncology (n = 19, 5%), Gastroenterology (n = 52, 14%), Primary Care (n = 29, 8%), and Obstetrics and Gynecology (OB/GYN) (n = 97, 27%). All identified physicians were surveyed. Respondents were assured of the anonymity of their responses.

### Survey

The survey consisted of 5 sections: (1) demographic information about the physicians surveyed, (2) awareness of and attitudes toward genetic testing, (3) opinions regarding genetic counseling, (4) knowledge regarding Mendelian genetics and interpretation of genetic test results, and (5) interest in learning more about genetic testing and counseling. Space on the questionnaire was provided for the physicians to comment on their responses. Multiple choice questions and Likert response formats were used to obtain the requested information. Responses were not mutually exclusive and, therefore, some questions allowed for more than one response. The survey is available upon request.

Members of the Department of Human Genetics at Mount Sinai critically reviewed the survey to verify content validity and ensure correct keying of knowledge questions. In August of 1996, the surveys were distributed to the 363 physicians. Prepaid, pre-addressed return envelopes were included with the surveys, but no incentives for completing the survey were offered. A second mailing of the instrument was sent out 8 weeks later. Due to the anonymous nature of the study, all physicians received a sec-

ond survey and were asked not to return a new survey if they had replied to the initial mailing.

### Analysis

Calculations were performed using Stata versions 5.0 and 6.0 (Stata Corp., College Station, TX). Comparisons between groups were made using analysis of variance when the dependent variable had a reasonably normal distribution. Comparisons between groups of variables with skewed distribution were made using the Mann-Whitney U-Test. Correlations between variables were assessed using the Spearman or Pearson correlation coefficients for ordinal and interval-level measures, respectively. When searching for comparisons between the "academic" and "private practice" groups, we excluded from those analyses physicians who failed to identify themselves as exclusively in either group (11 of 86 respondents provided either multiple responses or "other").

### Results

Of the 363 physicians initially surveyed, 91 (25%) returned surveys. Not all respondents answered every question. Twenty-two (22) of the 363 physicians were deemed ineligible for the study due to: (1) old or incorrect mailing address for which a new address was unavailable, (2) little patient contact (e.g., mostly research oriented) as noted on the returned surveys, or (3) inability to complete the survey due to unforeseeable events (e.g., death). Therefore, 89 (26%) returned surveys from the 341 eligible physicians were utilized for our analysis.

### Demographics

The mean age of those surveyed was 50, with the majority of respondents being male (74%). Of 88 respondents who answered the question, 28 (32%) had received information regarding genetic testing from commercial companies. A full breakdown of the results is found in Table 1.

### Education

Of the 89 respondents, 51 (57%) had taken a formal course in genetics at some point during their training, with 44 (86%) of those having taken a course in medical school. Time since completion of medical training was found to be a significant indicator of whether one had taken a formal course ( $p = 0.0006$ ). Those doctors who

**Table 1**  
*Demographic Data of Responding Physicians*

Age: n	Range (yrs)	Mean (yrs)
82	30–82	50 ± 13
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COT: n	Range (yrs)	
14	0–5	
19	6–10	
22	11–20	
15	21–30	
16	31+	
Total: 86	Mean: 18 ± 13	
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Sex: n = 86		
Male:	64 (74)*	
Female:	22 (26)	
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Professional Activity	n	Specialty
		n (%) Response Rate Within Each Field
Private:		
Primary Care	7	Cardiology 6 (16)*
Specialist	29	Geriatrics 8 (24)
Academic:		Gastroenterology 12 (23)
Primary Care	11	Neurology 14 (16)
Specialist	31	OB/GYN 14 (14)
Multiple <sup>o</sup> :	8	Oncology 2 (11)
Other <sup>†</sup> :	3	Pulmonary 1 (8)
Total:	89	No Response 8
		Total: 67

Received information regarding genetic tests: n = 88

Yes: 28 (32)

No: 60 (68)

\* Numbers within parentheses represent percentage of those responding

COT = Years since completion of training

<sup>o</sup> Multiple: These physicians identified themselves in more than one of the professional categories; 7 of 8 identified themselves as specialists + an additional category and were included when determining respondent's specialty.

<sup>†</sup> Other: These physicians were not used when analyzing "academic" and "private" groups.

had taken a course had completed training more recently (mean 13.7 years) than those who had not (mean 24.2 years). Only 33 (37%) of the 89 respondents, however, were currently reading articles concerning genetic testing, with journals being the most common source, as seen in Table 2.

### Self-Assessment

When asked how they would rate their knowledge about genetics and genetic testing, using a 4-point scale (1 = excellent, 4 = poor), 25 (29%) of the 87 respondents to this question rated it as excellent-to-good, while 62 (71%) rated it as fair-to-poor. Those physicians who had taken a formal course in genetics rated their knowledge higher than those who had not (mean of 2.7 vs.

**Table 2**  
*Educational Exposure*

Have taken a formal course in genetics: n = 89	
Yes:	51 (57)*
No:	38 (43)
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Stage of Training <sup>o</sup> :	n = 51
Medical School:	44 (86)
Residency:	2 (4)
Other:	10 (20)
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Read articles regarding testing on a regular basis: n = 89	
Yes:	33 (37)
No:	56 (63)
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Source <sup>o</sup> :	n = 33
Journals:	31 (94)
Newspaper:	15 (45)
Other:	6 (18)

\* Numbers within parentheses represent percentage of respondents.

<sup>o</sup> More than one response allowed for the question.

3.2;  $p = 0.01$ ), as did those who stated that they currently read articles regarding genetic testing (2.4 vs. 3.2;  $p < 0.0005$ ). There was no statistically significant difference between "academic" and "private practice" physicians as to how they rated their knowledge. Years since completion of training also was not associated with self-rated knowledge.

### Attitudes and Opinions Toward Testing

Using a 5-point scale, physicians were able to express their opinions about ordering genetic tests for various scenarios. These results are found in Table 3. While screening targeted populations for the identification of carriers of autosomal recessive disorders like Tay-Sachs was largely agreeable to physicians (81% agreed or strongly agreed), screening populations for carriers of similar disorders for the purpose of employability or health insurance coverage was found to be disagreeable (95%). Other scenarios, however, were not as easily delineated. Ordering prenatal testing of fetuses for an untreatable disease which would not manifest itself until later in life was agreeable to 44% of the respondents and disagreeable to 40%.

### Knowledge

Three categories of knowledge were assessed through our survey. The awareness of genetic tests available at the time of survey distribution (KN1) was determined utilizing the 12 disorders seen in Table 4. Overall, the results yielded a

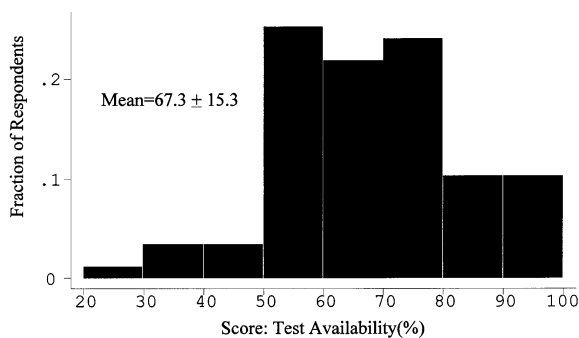
**Table 3**  
*Physician Attitudes Regarding Genetic Testing*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	n
A.	35 (41)*	37 (44)	7 (8)	1 (1)	5 (6)	85
B.	4 (4)	18 (22)	19 (23)	22 (27)	20 (24)	83
C.	17 (20)	25 (30)	14 (17)	18 (21)	10 (12)	84
D.	12 (14)	22 (26)	14 (17)	26 (31)	10 (12)	84
E.	34 (40)	35 (41)	6 (7)	6 (7)	4 (5)	85
F.	1 (1)	1 (1)	2 (2)	10 (12)	69 (83)	83
G.	7 (8)	9 (11)	11 (13)	26 (31)	31 (37)	84
H.	16 (19)	18 (21)	13 (15)	19 (23)	18 (21)	84
I.	27 (32)	28 (33)	11 (13)	10 (12)	9 (11)	85
J.	16 (19)	17 (20)	18 (22)	19 (23)	13 (16)	83

\* Numbers within parentheses represent percentage of respondents for each category.

- A. Presymptomatic screening of individuals at risk for *treatable* diseases such as hereditary nonpolyposis colon cancer, which may not manifest until later in life.
- B. Presymptomatic screening of individuals at risk for untreatable diseases such as spinocerebellar ataxia type 1 (SCA1), which may not manifest until late in life.
- C. General population, presymptomatic screening that identifies individuals at risk for common, treatable diseases such as hypertension.
- D. General population, presymptomatic screening that identifies individuals at risk for serious diseases such as breast cancer, though the results are not 100% definitive.
- E. Targeted population screening for the identification of carriers of autosomal recessive disorders, such as Tay-Sachs, to determine reproductive options.
- F. Population screening for the identification of carriers of autosomal recessive disorders to determine employability or health insurance coverage.
- G. Prenatal testing of fetuses at risk for treatable disease which may not manifest until late in life.
- H. Prenatal testing of fetuses at risk for untreatable disease which may not manifest until late in life.
- I. Prenatal testing of fetuses at risk for being born with a treatable, chronic disease such as Gaucher disease.
- J. Prenatal testing for fetuses at risk for being carriers or recessive or X-linked disorders.

bell-shaped distribution as shown in Fig. 1. The mean score was 67%, with no physician responding to all 12 disorders correctly. The second set of questions sought to assess physician understanding of Mendelian genetics (KN2). Six multiple choice questions, related to a genetic testing laboratory report which was provided, tested physicians on their understanding of genetic terms (e.g., heterozygosity and penetrance) and inheritance risks. Responses yielded a bimodal curve as seen in Fig. 2, with a mean score of 64%. The final question set sought to determine physician under-



**Fig. 1.** Genetic test availability correctly identified by physicians for various diseases (KN1).

standing of the mechanics behind and the proper interpretation of genetic test results (KN3). Two multi-answer, multiple choice questions assessed an understanding of the limitations of genetic testing and of the potential utility of the test results. Partial credit was given for these multi-answer questions. Responses produced a mean score of 65% and a bell-shaped distribution skewed rightward, as seen in Fig. 3. There was no significant correlation between scoring well in KN1 and scoring well in the other two knowledge sets. Physicians who identified themselves as practicing in an “academic” setting did significantly better in sets KN2 and KN3 ( $p = 0.002$  and  $0.008$ , respectively) when compared to “private practice” physicians. There was no statistically significant association between those achieving higher scores in the knowledge sets and those physicians who either had recently completed training or taken a formal course in genetics, or who currently read articles regarding genetic testing.

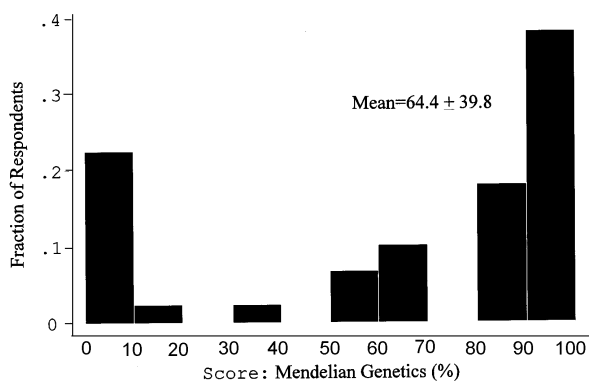
## Counseling

As seen in Table 5, 83 (95%) of 87 respondents believed that the doctor had a responsibility

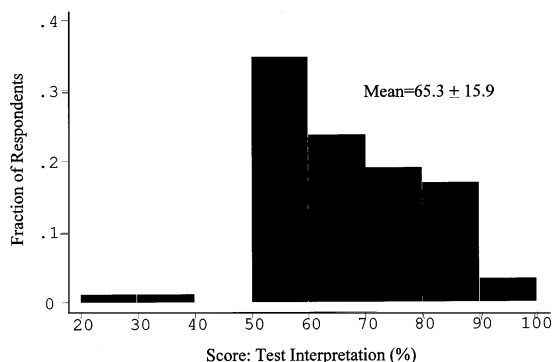
**Table 4**  
*Knowledge of Genetic Test Availability (KN1)*  
 n = 87

Disease	Available	Correct	Incorrect
Alzheimer's Disease	Yes	31 (36)*	56 (64)
Familial Breast Cancer	Yes	67 (77)	20 (23)
Sickle Cell Anemia	Yes	68 (78)	19 (22)
Huntington's Disease	Yes	78 (90)	9 (10)
Fragile-X Retardation	Yes	62 (71)	25 (29)
Diabetes	No	80 (92)	7 (8)
Cystic Fibrosis	Yes	73 (84)	14 (16)
Alcoholism	No	82 (94)	5 (6)
Myotonic Dystrophy	Yes	54 (62)	33 (38)
Hemophilia	Yes	56 (64)	31 (36)
Hypercholesterolemia	Yes	15 (17)	72 (83)
Charcot-Marie Tooth Disease	Yes	38 (44)	49 (56)

\* Numbers within parentheses represent percentage of respondents for each category.



**Fig. 2.** Correctly answered questions by physicians regarding Mendelian genetics (KN2).



**Fig. 3.** Correctly answered questions by physicians regarding the mechanics behind and the proper interpretation of genetic test results (KN3).

to provide the patient with counseling. The answer "doctors" was followed by "genetic counselors" (83%), "nurses" (36%), and "social work-

ers" (35%). Only 43 (51%) of 84 respondents, however, believed they actually had the time to discuss genetic counseling issues with their patients. Of 88 respondents, 86 (98%) stated that they would refer a patient for genetic counseling. While 80 (91%) of 88 respondents were aware of genetic counseling services in general, only 62 (71%) of them were aware of such counseling services at the major New York medical centers. And of the 80 physicians who were aware of counseling services, 42 (53%) had referred a patient for these services, with their impressions of the referral found also in Table 5.

### Further Information

Of 71 respondents, 48 (68%) were interested in learning more about genetic testing and counseling. Six choices regarding the form in which such information should be offered to physicians (e.g., CD-ROM, audio tapes, seminars) were presented for ranking according to preference. Since no one modality was found to be substantially more preferred than the others, the results of this analysis are not presented in this paper.

### Discussion

Our response rate of 26% was below our expectations and below the rates achieved by other study groups that had distributed surveys regarding similar topics (3, 5, 6, 8). Our study, however, unlike most of the other studies reviewed, did not provide incentives (i.e., continuing medical education [CME] credit or financial reimbursement), and our survey was also considerably longer than others. The study of Texas primary care physicians by Friedman et al. (2) had a response rate of 30%, similar to our own rate, without reimbursement. Nevertheless, the number and distribution of our responses were sufficient for analysis.

The majority of our respondents were male and had completed training less than 20 years prior to the study. Furthermore, most respondents identified themselves as specialists. This response reflects the high degree of specialization found at tertiary care hospitals such as Mount Sinai.

Those physicians who had completed training more recently were more likely to have had a formal course in genetics. This was also seen in a study by Hutter et al., in Ontario, Canada (8). Those who identified themselves as currently reading articles regarding testing or as having taken a formal course in genetics rated their

**Table 5**  
*Attitudes and Opinions Regarding Counseling*

Responsibility to provide the patient with counseling <sup>1</sup> : n = 87			
Doctor	83 (95)*		
Genetic Counselor	72 (83)		
Nurse	31 (36)		
Social Worker	30 (35)		
Receptionist	0 (0)		
Other	5 (6)		
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Genetic Counseling	Yes	No	n
Have time to counsel patients:	43 (51)	41 (49)	84
Aware of health professionals known as genetic counselors:	80 (91)	8 (9)	88
Aware of genetic counselors in the New York Area:	62 (71)	25 (29)	87
Would refer a patient to a genetic counselor:	86 (98)	2 (2)	88
Have referred a patient to a genetic counselor:	42 (53)	38 (47)	80
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Satisfaction with the referral: n = 41			
Mostly satisfied:	19 (46)		
Very satisfied:	15 (37)		
Neither:	5 (12)		
Mostly dissatisfied:	2 (5)		
Very dissatisfied:	0		

<sup>1</sup> More than one response allowed for the question.

\* Numbers in parentheses represent percentage of respondents for each category.

knowledge significantly higher than the other physicians, but did not score significantly higher on the three knowledge sets. The above results could imply that: (1) the issues considered important in our survey are not reflected or addressed in the articles read or courses taken by the surveyed physicians; (2) the courses these physicians took or the articles they read were inadequate for the genetic issues facing physicians today; or (3) since completing their course, the physicians had not kept up with the technological advances in genetics on a regular basis. Primary sources of information for physicians regarding genetics have been shown to include: journals (3, 5, 7, 8), genetics departments (8), colleagues (7, 8), and CME courses and meetings (5, 7). Utilization of most of these sources by Mount Sinai physicians was not assessed in our survey.

The attitudes and opinions expressed about the various scenarios related to genetic testing were fairly consistent with those found in other studies (6, 8). A large majority of physicians in our study failed to support testing for the purpose of employability or health insurance coverage. Other, more controversial issues generated greater division in the responses obtained. Those physicians more likely to counsel patients on such tests themselves are also more likely to be comfortable with ambiguity and confident in their ability to communicate the nature of these tests to their

patients (6). Of course, neither controversy nor uncertainty (among physicians or informed lay persons) has stopped numerous clinical laboratories from offering genetic testing to physicians and their patients. So it is only with caution, a solid base of knowledge, and appropriate counseling that a decision as to whether or not to undergo such tests can be properly made.

The low self-rating of knowledge regarding genetics and genetic testing implies that the responding physicians are aware of their limitations regarding the field of genetics. The mean scores for all three knowledge sets were over 60%, similar to findings in other studies assessing physician knowledge (3, 7). Such results, however, must be more closely examined. KN2 had a large standard deviation and produced a bimodal distribution. Physicians scored either very well or very poorly in this section, implying that they had either a satisfactory or an insufficient knowledge of Mendelian genetics. Furthermore, the rightward skew of KN3 implies that although there was a large number of respondents scoring around the mean, there was also a large range of respondent scores on the higher end of the grading scale. The fact that scoring higher in KN1 did not correlate with scoring higher in KN2 or KN3 was also of interest, implying that those physicians who were aware of the availability of various genetic tests

were not necessarily able to properly identify those patients at risk and, furthermore, did not necessarily understand the limitations of such tests. Conversely, those physicians who displayed a significantly better understanding of Mendelian inheritance patterns or were aware of the limitations of testing (e.g., those physicians practicing in an "academic" setting) were not necessarily aware of the tests they could be offering their at-risk patients. Either way, patients would *not* be properly informed, either because of a lack of understanding of the tests or because of a lack of awareness of such tests by their physicians. Such a finding was seen in studies regarding colon and breast cancer testing, which showed that patients were referred for testing by their physicians even though the patients failed to meet the recommended criteria and guidelines for the genetic tests (5, 9). The finding that "academic" physicians scored higher than "private practice" physicians in KN2 and KN3 was also true of the survey of almost 1800 physicians by Hofman et al. (7), which showed full-time faculty scoring higher on their knowledge questions than did other non-geneticist physicians. This may be due to the fact that "academic" physicians may have better access to this information through colleagues or nearby departments; however, cannot be determined from the results of this study.

Patient response to genetic test results can be quite varied and can have long-term impact (10–12). With the rapid increase in technology, coupled with the insufficient number of geneticists and genetic counselors, it may be the patient's primary physician who will have to shoulder the responsibility of genetic counseling (13). (Nearly one-third of those responding had already been sent information from a commercial company offering genetic testing.) Furthermore, it is these physicians who will have the duty to help the patient deal with the consequences of the test results, both in terms of offering support and providing preventive measures (4, 13, 14). Genetic testing is ethically difficult because the results affect not only the patient being tested, but the patient's entire family (4, 13). Issues regarding who else can or should be informed about a *potential* predisposition to a disease will be difficult to deal with, especially considering the lack of cures or even agreed-upon treatments for a variety of these diseases (4, 13). While a vast majority of our respondents believed the physician had some responsibility for counseling the patient, only 51% believed they actually had sufficient time. Furthermore, counseling may not be

adequate or appropriately nondirective in nature, if given by physicians who are inadequately trained in dealing with these issues (6). While nearly all the respondents would have no difficulty in referring a patient to a genetic counselor, only 71% of them were aware of the services in the New York area. Of those aware of geneticists and genetic counselors, more than half had utilized their services, implying that the demand for such services exists, with most physicians mostly-to-very satisfied with the results.

Of those responding, more than two-thirds expressed interest in learning more about genetic counseling and testing. Unfortunately, no preferred modality for distributing this information was found through our survey, and further research into this is needed.

There are some inherent limitations in our study. The survey was quite lengthy (7 pages) and, due to our own financial and resource limitations, we were not able to offer any incentives to those surveyed. The length, unfortunately, caused many to return only partially completed surveys. While this did not affect most parts of our survey, the final section, in which we inquired about learning more about genetics, was left incomplete more frequently than other sections ( $n = 71$  vs.  $n > 82$  responding in other sections). Nevertheless, a large majority of those responding to this section did request more information. The low response rate overall, despite its distribution among the various medical fields, may not adequately assess the entire population surveyed. Responses may be biased, as respondents to this lengthy survey may have a stronger interest in genetics, whether due to personal or professional reasons. The small number of respondents also weakened our ability to break down our data further. There were too few physicians to make any significant conclusions about each individual specialty. Though pediatricians are also affected by the new genetic technology, they were not included in this study, as we chose to focus on physicians dealing with adult-oriented diseases.

## Conclusions

With genetic technology rapidly changing our understanding of disease and our approach to treatment, it is a challenge for physicians to keep up. There is a need for further education to help physicians become aware of currently available tests, and to help them properly convey the risks and benefits of such tests to their patients. Awareness of the available counseling services

needs to be heightened to optimally provide physicians and their patients with the resources and services they desire. Finally, the most effective and efficient methods for providing this information need to be determined.

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