Incorporation of Genetics in Primary Care Practice

Will Physicians Do the Counseling and Will They Be Directive?

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Objective: To determine, by response to a scenario, how willing primary care physicians would be to counsel a couple about prenatal diagnosis of cystic fibrosis and how directive they would be about whether the couple should undergo prenatal diagnosis and whether the couple should terminate the pregnancy if the fetus is affected.

Design: Survey of a random sample of primary care physicians, psychiatrists, and genetics professionals in 10 geographically representative states.

Respondents: Sixty-five percent (N=1140) of 1759 obstetricians, pediatricians, internists, family practitioners, and psychiatrists, and 79% (N=280) of medical geneticists and genetic counselors.

Outcomes and Results: Respondents were evenly divided on whether they would counsel about prenatal diagnosis or refer to a genetic counselor (49.4% and 50.6%, respectively). Those who indicated that they would counsel were likely to have greater knowledge about genetics, greater confidence in communicating about genetics, and higher tolerance for ambiguity and were more likely to have completed their medical training since 1971 and to practice in a rural area. Forty-four percent of physicians would give an opinion about prenatal diagnosis. Men would be more likely to give an opinion than women ($P<.005$). Only 9.6% of respondents would give an opinion regarding abortion. These respondents were more likely to come from specialties with less exposure to genetics and to value attendance at religious services. Primary care physicians were more likely to give their opinions about prenatal diagnosis and abortion than genetics professionals.

Conclusions: To the extent that attitudes are reflected in practice, genetic counseling may be more directive when provided by primary care physicians than by genetics professionals, unless primary care physicians' growing involvement in genetics changes their attitudes.

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With the proliferation of new genetic tests that will detect carriers of disease-causing genes and predisposition to disease and will diagnose disease presymptomatically, there will be an insufficient number of trained genetics professionals to offer testing.[1] Consequently, genetic testing is increasingly likely to become a part of primary care practice.[2,3] To adapt to such rapid advances in genetic information and diagnosis, physicians will have to examine their role in making this information available to their patients at several stages of decision making. The incorporation of genetic testing into clinical medicine is currently in a transitional phase, where the information about the advantages and disadvantages of genetic tests is still incomplete and professional norms have not yet evolved to take into account new developments.[4]

Little is known about how primary care physicians will incorporate new genetic technologies into their practices. Some might resist offering their patients genetic services for various reasons: they lack experience and confidence in discussing genetics; they feel burdened and overwhelmed by what seems like an increasing list of obligations in the face of decreasing time and resources; or they

See Methods on next page

METHODS

STUDY DESIGN AND POPULATION

An equal number of primary care physicians (family physicians, internists, obstetrician-gynecologists [hereafter referred to as obstetricians], and pediatricians) and psychiatrists were selected at random from the American Medical Association Physician Masterfile for each of the following states: New York, Illinois, Nebraska, Pennsylvania, Utah, Maine, Oregon, California, South Carolina, and Texas. (Each of these states was selected at random from the 10 Genetics Network Regions established by the Health Resources Administration–Department of Health and Human Services). Because the overall study was intended to focus on what has *recently* been taught in medical schools, physicians (the term *physicians* will be used to refer to primary care physicians *and* psychiatrists) within each specialty who graduated between 1971 and 1985 were sampled at a ratio of 2:1 to those who graduated between 1950 and 1970. Only members or candidate members of the following societies (as determined by a search of their directories) were retained in the sample: American Academy of Family Physicians (AAFP), American College of Physicians (ACP), American College of Obstetricians and Gynecologists (ACOG), American Academy of Pediatrics (AAP), and American Psychiatric Association (APA). They are predominantly board-certified. To validate the survey instrument and for comparison, medical geneticists and genetic counselors were included in the study and sampled from membership lists provided by the American Board of Medical Genetics. A detailed description of survey methods appears elsewhere.[8]

PROCEDURES

A questionnaire was mailed to all eligible physicians with a promise that we would not link names and individual responses. In an effort to maximize our response rate, physicians were offered $25 and continuing medical education credits for completing the questionnaire. In addition, endorsements were obtained from the relevant medical specialty societies and from local chapter heads of three of the societies. Follow-up included postcard reminders, questionnaire remailings, and phone calls.

CONSTRUCTION OF VARIABLES

All items in this component of the questionnaire were in multiple-choice format. The instructions for responding to the CF scenario conceded that some of the questions might not be relevant to the respondent's specialty but encouraged all respondents to complete the entire scenario. The scenario presented physicians with the following three hypothetical situations:

OUTCOME VARIABLES

1. Referral for Genetic Counseling

Respondents were told to imagine a pregnant couple coming to them, both carriers of the gene for CF. Respondents then were asked whether they would "discuss prenatal diagnosis with the couple before (or instead of) referring them to a genetic counselor" or "immediately refer them to a genetic counselor." This question was not presented in a general way. It was preceded in the scenario by several questions that were very specific to this couple's situation and allowed the respondent to assess his or her own knowledge level about this couple's risk. Therefore, by the time respondents reached this question, they would be quite clear about whether they would feel capable of handling the case themselves or would make a referral. Regardless of whether they said they would discuss or refer, respondents were then told that the couple returned to them to decide about prenatal diagnosis.

2. Opinion Regarding Prenatal Diagnosis

Respondents then were asked whether, in discussing the pros and cons of prenatal diagnosis with this pregnant carrier couple, they would "give their opinion about whether it is a good idea but emphasize that they have to make their own decision" or "refrain from expressing an opinion or giving advice and emphasize that they have to make their own decision" as to whether the woman should undergo prenatal diagnosis.

3. Opinion Regarding Abortion

Respondents then were asked to imagine that this same couple has prenatal diagnosis and that the fetus has CF. Respondents were asked what kind of opinion they would give the

have ethical concerns about the value and implications of genetic testing. Among those who might welcome the opportunity to incorporate genetic services into their practices, it is not known whether or to what extent they would be nondirective in interactions with patients. Nondirectiveness certainly has not been as much of a focus of primary care training as it has been of genetics training. Moreover, despite the belief among most genetics professionals that genetic counseling should be nondirective,[5] the desirability and practicality of nondirectiveness is currently being challenged even in genetics circles.[6,7]

As part of a larger descriptive survey conducted in 1991 to assess physicians' and genetics professionals' knowledge of genetics and genetic testing, we investigated physicians' perceptions of their role in prenatal genetic counseling, using cystic fibrosis (CF) as a case study. We were interested in how willing physicians would be to discuss prenatal diagnosis of CF with a couple found to be carriers of the CF gene and how directive they would be when discussing whether the couple should undergo prenatal diagnosis and whether they should terminate the pregnancy if the fetus is affected. In addition, we were interested in how physicians' specialty, as well as their knowledge of genetics, confidence in communicating about genetics, tolerance for ambiguity, and

couple about terminating the pregnancy. Choices included (1) "recommend that the mother continue the pregnancy because of (*a*) moral objection to abortion or (*b*) the improved outlook for a child with CF born today," (2) "recommend that the mother terminate the pregnancy because of (*a*) the burden that a child with CF will be to the child and the family or (*b*) the burden that a child with CF will be to society," and (3) "make no recommendation, review the issues and options with the couple, and indicate that they must decide whether to continue or terminate the pregnancy." Responses were divided into those in which an opinion of any kind would be given vs those in which no opinion would be given. Both opinion variables (regarding prenatal diagnosis and abortion) were taken as measures of directiveness in communication.

PREDICTOR VARIABLES

Specialty

In addition to sampling four primary care specialties, psychiatrists were included because of their potential involvement in genetic testing, eg, for susceptibility to major affective disorders. For comparison purposes, medical geneticists and genetic counselors also were included in the sample. Genetics professionals were instructed to skip the question about "referral."

Knowledge

Knowledge scores were derived from the number of correct responses to 26 knowledge questions. A detailed description of the development and results of the knowledge test appears elsewhere.[9]

Tolerance for Ambiguity

A previously validated seven-item scale was used to measure people's general reaction to ambiguous situations. Respondents were asked to rate the extent to which they agreed or disagreed with each item. Overall tolerance for ambiguity scores were then created by summing the responses. The reliability; based on Cronbach's α test, was .75. A detailed description of the psychometric analyses involved in scale development appears elsewhere.[10]

Confidence

A scale of confidence in communicating about genetic testing was developed, modeled after previous work.[11-13] For each of nine items, respondents were asked to check off one of six response categories, ranging from "not at all confident" to "very confident." A total confidence score was created by summing responses to the individual items. The reliability of the scale was .89 (Cronbach's α test).

Attendance at Religious Services

Respondents were asked the extent to which it was important to attend religious services regularly, using a six-point scale ranging from "very unimportant" to "very important." Based on the frequency distribution of responses, a dichotomous variable then was created for subsequent analysis (those who said "very important" vs everyone else).

Year of Graduation

Consistent with the decision we made to oversample recent graduates from medical school, we dichotomized year of graduation into those who graduated from 1971 through 1985 and those who graduated prior to 1971.

Geographic Area

Respondents were grouped into those who reported practicing in a rural area vs elsewhere (urban, suburban, or small city).

ANALYSIS

Knowledge, confidence, and tolerance for ambiguity scores were dichotomized according to the frequency distributions of the respective scores, with the 50th percentile falling in the upper group. Physicians in the higher knowledge group had scores of at least 75% correct. Once all variables were in categorical form, χ^2 tests of association were used to look at the relationship between outcome and predictor variables. Stepwise logistic regression was used to adjust for confounding effects of the several characteristics associated with referring to a genetic counselor and giving opinions about pregnancy termination. All analyses were done using SPSS/PC+.

demographic characteristics, contributed to variations in the likelihood that they would discuss prenatal diagnosis and be directive in their discussions.

RESULTS

RESPONSE RATES

In calculating response rates, questionnaires that did not reach the intended respondent because of death, retirement, or relocation were not included. The response rate from medical geneticists and genetic counselors was 79.1%. The response rate from physicians was 64.8%, ranging from 61% for obstetricians to 72.6% for pediatricians. Respondents were more likely to be younger than 40 years and to have graduated from medical school after 1977. A detailed analysis of differences between respondents and nonrespondents appears elsewhere.[8]

PHYSICIANS' PERCEPTIONS OF THEIR ROLE IN PRENATAL GENETIC COUNSELING

With respect to the three outcome variables, physicians who responded were evenly divided as to whether they

Table 1. Directiveness of Physicians vs Genetics Professionals in Prenatal Genetic Counseling

Measure of Directiveness	No. Responding (% Responding Yes)		
	Primary Care Physicians	Medical Geneticists	Genetic Counselors
Would give their opinion as to whether the woman should undergo prenatal diagnosis (vs refraining from giving their opinion)	1113 (43.8)*	141 (16.3)	139 (2.9)
Would give their opinion about pregnancy termination to a mother whose fetus was diagnosed with cystic fibrosis (vs refraining from giving their opinion)	1137 (9.6)†	141 (0.7)	139 (1.4)

**All three groups were significantly different from each other at P<.0001.*
†Physicians were different from genetics professionals at P<.0001, but medical geneticists and genetic counselors did not differ from each other.

would discuss prenatal diagnosis with a carrier couple (49.4%) or refer them immediately to a genetic counselor (50.6%). Physicians were almost evenly divided in their willingness to give an opinion as to whether this carrier couple should undergo prenatal diagnosis (43.8% said yes, 56.2% said no). Surprisingly, there was no association between willingness to discuss prenatal diagnosis and willingness to give an opinion about prenatal diagnosis. That is, physicians who would discuss prenatal diagnosis were no more likely to give their opinions than those who would refer for prenatal diagnosis. Very few physicians (9.6%) indicated that they would give their opinions regarding pregnancy termination. Compared with either group of genetics professionals, physicians were significantly more likely to give their opinions about prenatal diagnosis; medical geneticists were more likely than genetic counselors to give their opinions (**Table 1**).

Physicians' willingness to discuss prenatal diagnosis and give their opinions differed by specialty. **Table 2** shows that obstetricians would be more likely than other specialists to refer a couple for prenatal genetic counseling. Family practitioners would be more likely than obstetricians to discuss rather than refer for prenatal diagnosis. Psychiatrists would be the least likely to give an opinion about prenatal diagnosis, although this difference was not statistically significant. Compared with the other specialists, family practitioners and internists would be more likely to give their opinions about abortion.

CHARACTERISTICS ASSOCIATED WITH PHYSICIANS' PERCEPTIONS OF THEIR ROLE

As shown in **Table 3**, physicians who indicated that they would discuss prenatal diagnosis for CF with a carrier couple rather than refer the couple immediately to a genetic counselor were likely to have greater knowledge about genetics, greater confidence in communicating about genetics, and higher tolerance for ambiguity and were more likely to have graduated recently from medical school and to practice in a rural area. Gender and attendance at religious services were not associated with making referrals. We also determined that rural and urban physicians did not differ in their knowledge, confidence, tolerance for ambiguity, and year of graduation.

With respect to the likelihood that respondents would give their opinion about whether a couple should undergo prenatal diagnosis, gender was the only significant variable. Women were less likely than men to report that they would give an opinion. This was particularly true for female obstetricians (χ^2=8.5, $P<.005$) and psychiatrists (χ^2=18.7, $P<.00005$). There was no gender difference among pediatricians.

A relatively small percentage of respondents indicated that they would give an opinion regarding pregnancy termination. Of those who would offer an opinion (N=112), 58.9% would object to termination and 41.1% would recommend termination. Those offering an opinion were more likely to have low knowledge scores, low confidence scores, and low tolerance for ambiguity and were more likely to be male, to have graduated from medical school prior to 1971, and to

Table 2. Physicians' Perceptions of Their Role in Prenatal Genetic Counseling by Specialty

Specialty	No. of Physicians	Would Discuss Prenatal Diagnosis, %		Would Give an Opinion About Prenatal Diagnosis, %		Would Give an Opinion About Abortion, %	
		Discuss	Refer*	Yes	No*	Yes	No
Pediatricians	257	54.7	45.3	44.4	55.6	5.8	94.2
Family practitioners	211	56.4	43.6	42.6	57.4	16.4	83.6
Obstetricians	224	36.9	63.1	44.7	55.3	5.4	94.6
Internists	214	48.1	51.9	50.5	59.5	14.5	85.5
Psychiatrists	230	50.4	49.6	37.0	63.0	7.0	93.0
Total	**1136**	49.4	50.6	43.8	56.2	9.6	90.4

**P<.0003 by overall χ^2 analysis.*

Table 3. Factors Associated With Physicians' Perceptions of Their Role in Prenatal Genetic Counseling

	No. of Physicians	Would Discuss Prenatal Diagnosis, %		Would Give an Opinion About Prenatal Diagnosis, %		Would Give an Opinion About Abortion, %	
		Discuss	Refer	Yes	No	Yes	No
Knowledge							
High	586	55.8*	44.2	42.9	57.1	6.6*	93.4
Low	543	42.5	57.5	44.7	55.3	12.7	87.3
Confidence							
High	629	53.1†	46.9	43.3	56.7	7.1†	92.9
Low	500	44.8	55.2	44.3	55.7	12.6	87.4
Tolerance for ambiguity							
High	536	53.4‡	46.6	44.3	55.7	7.8‡	92.2
Low	593	45.9	54.1	43.2	56.8	11.2	88.8
Gender							
Male	915	49.5	50.5	47.0†	53.0	10.7†	89.3
Female	222	49.1	50.9	30.6	69.4	5.0	95.0
Year of graduation							
1950-1970	365	38.4†	61.6	46.9	53.1	14.1†	85.9
1971-1985	764	54.7	45.3	42.3	57.7	7.4	92.6
Geographic area							
Rural	92	67.4*	32.6	50.5	49.5	11.8	88.2
Nonrural	1031	48.0	52.0	43.2	56.8	9.2	90.8
Religiosity							
Very important	230	47.8	52.2	41.7	58.3	20.6†	79.4
Not very important	890	49.9	50.1	44.3	55.7	6.8	93.2

*P<.0005.
†P<.005.
‡P <.05.

indicate that attending religious services regularly is very important.

Table 4 shows the multiple regression analyses used to predict whether physicians would discuss prenatal diagnosis with their patients and whether they would give their opinions regarding pregnancy termination. (Multivariate analysis was not performed to predict whether physicians would give an opinion about prenatal diagnosis since gender was the only predictor variable associated with this outcome.) In the first model, the strongest predictor of whether physicians would discuss prenatal diagnosis was practicing in a rural area, followed by recent graduation, high knowledge score, high tolerance for ambiguity, and specializing in family practice as opposed to obstetrics. In the second model, physicians who would give their opinions regarding abortion were 3½ times more likely to value attendance at religious services, over 2½ times more likely to be internists or family practitioners than obstetricians, and more likely to have low confidence scores and to have graduated prior to 1971.

Table 4. Logistic Regressions to Predict Physicians' Perceptions of Their Role in Discussing Prenatal Diagnosis and Giving Their Opinions About Abortion

	Logistic Regression Coefficient (SE)	Odds Ratio (95% Confidence Interval)	P
Discussing prenatal diagnosis			
High knowledge	.41 (.13)	1.5 (1.16-1.95)	.001
High tolerance for ambiguity	.30 (.12)	1.4 (1.06-1.72)	.01
Recent graduation	.53 (.14)	1.7 (1.28-2.25)	.0001
Rural setting	.65 (.24)	1.9 (1.19-3.09)	.008
Family practice*	.34 (.17)	1.4 (1.01-1.97)	.04
Giving their opinion about abortion*			
High confidence	−.54 (.21)	0.6 (0.38-0.72)	.01
Recent graduation	−.76 (.21)	0.5 (0.31-0.71)	.0004
Religiosity	1.3 (.22)	3.5 (2.36-5.7)	.00001
Family practice*	.95 (.25)	2.6 (1.57-4.26)	.0002
Internal medicine*	1.0 (.26)	2.8 (1.62-4.57)	.0001

*Obstetrics was the reference group for specialty in both regressions.

COMMENT

In this study we investigated primary care physicians' and psychiatrists' perceptions of their role in communicating about prenatal diagnosis for CF, a new genetic technology. Of major interest was the extent to which physicians would engage in genetic discussions (counsel) at all and would deviate from the tradition of nondirectiveness on which genetic counseling theoretically is based[14] and that was supported by the attitudes of genetics professionals in this study.

COUNSELING ABOUT PRENATAL DIAGNOSIS VS REFERRING FOR COUNSELING

The results show that half of physicians would be willing to counsel about prenatal genetic testing and half would prefer to leave such discussions to genetics professionals. To the extent that respondents who were willing to discuss prenatal testing had higher genetics knowledge scores, were better able to tolerate ambiguity, and were educated more recently (and, therefore, perhaps were better able to appreciate the increasing importance of genetics in medicine), they were better qualified to counsel than those who would refer. Whether those who would counsel were *adequately* qualified cannot be answered by this study.

Respondents' willingness to discuss prenatal diagnosis is influenced by their specialty and by whether they practice in a rural area. Obstetricians are more likely than other specialists to refer their patients for counseling about prenatal diagnosis, perhaps because they have established connections with genetic counselors as a way of managing the demand for genetic services in their practice. Obstetricians may begin to do more counseling as they perform more of their own amniocenteses. That family practitioners would be more likely than obstetricians to discuss prenatal diagnosis may be partially explained by their greater likelihood of practicing in rural areas ($P<.00001$). It is reassuring to know that physicians in this study who practice in remote locations appear no less qualified (as measured by knowledge, tolerance for ambiguity, and year of graduation) to offer genetic services than those in urban areas.

WILLINGNESS TO GIVE OPINIONS REGARDING PRENATAL DIAGNOSIS AND ABORTION

Physicians were significantly more willing than medical geneticists and genetic counselors to give their opinions about prenatal diagnosis and abortion, and male physicians were more likely to do so than female physicians. A similar gender difference was found among genetics professionals ($P<.00001$). That genetic counselors would be even less likely than medical geneticists to give an opinion about prenatal diagnosis could be related to the fact that genetic counselors are predominantly female, or it could be related to their nondirective training.

Among primary care specialists, almost half of obstetricians (the specialists most involved in prenatal diagnosis) reported a willingness to give an opinion about prenatal diagnosis. Obstetricians were the least likely to give their opinions about abortion. Physicians who would give their opinions about abortion, although a minority, were more likely to attend religious services regularly and to come from specialties with less exposure to genetics (eg, internal medicine). The directiveness of those with less exposure to genetics could reflect less experience with genetics or a failure to distinguish reproductive issues from other topics these physicians may discuss with their patients in a directive or authoritative manner. There is a need for further study of motivational differences between physicians who are willing to give their opinions and those who are not.

PHYSICIANS WHO give their opinions do not necessarily undermine autonomous decision making on the part of patients, especially if patients ask for their physician's opinion. Opinion seeking on the part of patients may occur more frequently in the context of long-term relationships with providers such as family practitioners than in the context of one-time contacts with genetics professionals. Many physicians and patients believe that it is appropriate and desirable for physicians to give advice to patients. Some believe that as long as physicians acknowledge their biases and uncertainties, their giving advice to patients is consistent with respect for patient autonomy unless the patient is unduly influenced. How and in what context physicians give their opinions may be of greater importance than whether they do so.

There is considerable controversy about when, if ever, it is appropriate to give opinions to patients when communicating about genetic testing. One view says that the practitioner's opinion should never be given in any context because genetic decisions are inherently value-laden and the perception of practitioners (especially male physicians[15]) as powerful can influence patients to behave in ways that are not consistent with their own values. This view provides the basis of value neutrality in genetic counseling. Another view says that giving opinions is inappropriate only in the context of reproductive genetic counseling, where decisions about conception and abortion are implicated. A third view says that the appropriateness of the practitioner's opinion regarding genetic testing depends on whether there is a beneficial treatment that can be offered. For example, when presymptomatic genetic testing for familial colon cancer is available, physicians would be remiss in not recommending testing, as colectomy can be beneficial to people who carry the gene. In such cases, genetic decisions are no different than other medical decisions in which physicians are expected to give their advice. In contrast, if no effective treatment is available (eg, Huntington's disease), it is difficult to justify recommending presymptomatic testing.

Although it is a matter of some debate whether the tradition of nondirectiveness espoused by genetics professionals is real and not rhetorical,[6] at the very least, value neutrality and nondirective counseling have been the prevailing "ideals." Our findings suggest that the ethos regarding patient counseling may be different for many primary care physicians, and as these physicians become more involved in genetic counseling, the extent of directiveness may change.

LIMITATIONS AND FUTURE RESEARCH

The validity and generalizability of these results may be affected by several limitations. First, there may be a tendency for respondents to provide socially desirable answers. Given that a substantial number of respondents admitted a tendency to give an opinion and that many more are likely to give their opinions without acknowledging it, our findings are likely to reflect a conservative estimate of the tendency of physicians to be directive.

Second, our findings may not be representative of all physicians. For example, the facts that we oversampled younger physicians, that younger physicians were more likely to respond, and that younger physicians appear more likely than older graduates to discuss prenatal diagnosis suggest that we might be overestimating the degree to which physicians in the general population would counsel about prenatal diagnosis. Similarly, the fact that younger physicians appear less likely than older graduates to give their opinions about abortion suggests that our findings may underestimate the degree to which physicians in the general population are directive about abortion. Younger physicians may be more aware of the sensitivity of the abortion issue or may be trained to be less directive in general than physicians in years past. Alternatively, young physicians may become more directive as they are in practice longer. These are empirical questions that warrant prospective study.

Third, this study is limited by the hypothetical nature of the scenario and the focus on CF. Both are likely to have put psychiatrists and internists at a disadvantage. These specialists have not had much experience in offering genetic services of any kind, least of all prenatal genetic testing. We are unable to generalize from the responses of psychiatrists and internists about how they might respond to the availability of DNA testing for common adult diseases, such as familial Alzheimer's disease and colon cancer, respectively. If the future demands that these specialists have a clear role in genetic testing for adult-onset diseases, some form of postgraduate training or continuing medical education may be needed to provide them with opportunities to learn about genetics and counseling.

Subsequent research should evaluate specialty- and gender-specific attitudes toward genetic interventions and should incorporate qualitative data-collection methods, such as individual interviews, focus groups, and audiotaped patient encounters. Such studies would provide a more accurate picture of how various primary care specialties are likely to respond to and communicate about new genetic tests and would enable us to assess the effect of other influences, such as their attitudes toward abortion. By achieving a better understanding of how physicians will incorporate new genetic technologies into primary care practice, we can enable appropriate, safe, effective, and equitable use of these technologies.

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