Psychiatric Genetics: A Survey of Psychiatrists' Knowledge, Opinions, and Practice Patterns

Christine T. Finn, M.D.; Marsha A. Wilcox, Sc.D., Ed.D.; Bruce R. Korf, M.D., Ph.D.; Deborah Blacker, M.D., Sc.D.; Stephanie R. Racette, M.A.; Pamela Sklar, M.D., Ph.D.; and Jordan W. Smoller, M.D., Sc.D.

Objective: Knowledge about the genetic basis of psychiatric illness is growing rapidly, and psychiatrists may be called upon to incorporate this information into clinical practice. The goal of this study was to assess psychiatrists' familiarity with and attitudes toward genetic information.

Method: We surveyed 844 participants, the majority of whom were psychiatrists, attending a continuing medical education course in the fall of 2002 and measured knowledge, opinions, and current practice patterns in regard to psychiatric genetics.

Results: Responses were received from 352 psychiatrists (54% of those surveyed). Most psychiatrists correctly answered fewer than half of survey items assessing general and psychiatric genetic knowledge. While 83% considered it their role to discuss genetic information with patients and families, fewer than 25% felt prepared or competent to do so. In response to hypothetical questions regarding genetic testing, a substantial proportion of psychiatrists indicated willingness to use such tests for diagnostic clarification, as well as presymptomatic and even prenatal risk prediction. The majority of respondents expressed interest in further genetics education.

Conclusions: Our results suggest that psychiatrists view genetic information as clinically relevant, but have limitations in knowledge that may impact the incorporation of psychiatric genetics into clinical practice.

(J Clin Psychiatry 2005;66:821-830)

Received Dec. 2, 2004; accepted Jan. 27, 2005. From the Department of Psychiatry (Drs. Finn, Blacker, Sklar, and Smoller and Ms. Racette), the Psychiatric Genetics Program in Mood and Anxiety Disorders (Drs. Finn and Smoller and Ms. Racette), and the Psychiatric and Neurodevelopmental Genetic Unit (Drs. Sklar and Smoller), Massachusetts General Hospital, Boston; Boston University Medical School Genetics Program, Boston, Mass. (Dr. Wilcox); University of Alabama at Birmingham, Birmingham (Dr. Korf); and Broad Institute, Boston, Mass. (Dr. Sklar).

A portion of this study was supported by the American Psychiatric Institute for Research and Education on Severe Mental Illness-Janssen Pharmaceuticals Research Scholarship (Drs. Finn and Smoller) and by grants from the National Institute of Mental Health (K-08 MH01770-02) (Dr. Smoller) and the National Institutes of Health (ELSI grant R01 HG01831) (Dr. Blacker).

The authors acknowledge the directors and staff of the Massachusetts General Hospital/Harvard Medical School psychopharmacology course for their assistance in completing this project: John B. Herman, M.D., Jerrold F. Rosenbaum, M.D., and Maurizio Fava, M.D., along with Gail Dickson, M.P.A. In addition, we thank Ming Tsuang, M.D., Ph.D., D.Sc., Stephen Faraone, Ph.D., Susan L. Santangelo, Sc.D., Robin Blatt, R.N., M.P.H., Beth Rosen-Sheidley, M.S., C.G.C., Holly Peay, M.S., C.G.C., and Catherine Bove, R.N., M.Ed., A.P.N.G., for their helpful feedback in the development of the survey. We also thank Mason Freeman, M.D., William Crowley, M.D., and Virginia Hughes, M.S., for their collaboration.

Corresponding author and reprints: Christine T. Finn, M.D., 77 Avenue Louis Pasteur, Suite 250, Boston, MA 02115 (e-mail: cfinn@partners.org).

substantial body of evidence over the past several decades has established that many psychiatric disorders can be familial and that genes play an important role in their etiology.¹ Family, twin, and adoption studies, in addition to demonstrating the familiality and heritability of a variety of psychiatric illnesses, have also provided empiric estimates of familial risk. At present, having a close family member affected with a mental illness (e.g., schizophrenia) is the greatest known risk factor for development of disease.² To date, the only common psychiatric disorder for which specific susceptibility genes have been conclusively identified is Alzheimer's disease,³ although recent reports suggest that solid findings are emerging for other disorders. For example, within the past 2 years, strong evidence implicating specific genes as risk factors for schizophrenia has been reported.⁴

As the genetic basis of psychiatric disorders is better understood, psychiatrists will need to integrate genetic information into clinical practice. At this point, it is unclear how well prepared psychiatrists are to meet this demand. Other fields of medicine—oncology, endocrinology, neurology—are facing similar challenges, as genetic ad-

vances have transformed the diagnosis and management of a number of common medical illnesses. To date, little is known about the potential clinical impact of genetics in psychiatry, although available evidence from the few studies that address these topics suggests strong interest in genetic counseling and genetic testing on the part of providers, patients, and families.⁵⁻¹⁰ For example, in 1 survey of 90 patients with bipolar disorder, 100% indicated that they would definitely or probably take a test to determine whether they were "carrying a gene for bipolar disorder" if it were available.8 In the coming years, psychiatrists will be challenged to acquire medically relevant genetic information and to use this knowledge to inform clinical decision making. There are relatively few studies that have examined general genetic concepts among physicians, but the available evidence suggests that there are gaps in knowledge regarding genetics among physicians from a variety of clinical specialties.^{11–14}

Incorporation of genetics into the clinical arena presents challenges and dilemmas beyond simply acquiring and interpreting new knowledge. For example, the traditional approach to genetic counseling emphasizes "nondirectiveness" (although there is controversy about whether completely nondirective counseling is possible or desirable).15-18 Previous studies suggest that most physicians may be more directive in counseling patients and families than are genetics professionals (e.g., medical geneticists, genetic counselors, and genetic nurses).¹⁹⁻²¹ Genetic counseling involving mental illness will raise ethically complex issues, and, if they become available, genetic tests for psychiatric disorders may magnify these issues for clinicians, patients, and families. At present, little is known about how psychiatrists view genetic testing for psychiatric disorders and under what circumstances they might favor its use.

Given the substantial impact that genetic research is likely to have on psychiatry in the coming decade,²² we sought to assess clinicians' familiarity with and opinions about genetics and its role in clinical practice. The current study was undertaken to examine psychiatrists' knowledge and attitudes regarding the use of genetic information. To our knowledge, this represents the largest such survey of psychiatrists, and the results highlight the potential value of additional genetic education for psychiatrists.

METHOD

Sample

The sample comprised all clinicians (N = 844) attending a continuing medical education (CME) course on psychopharmacology held in Boston, Mass., in the fall of 2002. Ninety-seven percent of total course participants were from the United States or Canada. The majority of course attendees were psychiatrists (77% of total course registrants). For these analyses, we examined only the responses of psychiatrists. All course participants received a copy of the survey as part of their course materials at registration. All surveys that were returned prior to the end of the first day of the course were eligible for inclusion in the analysis. In an effort to increase response rates, course directors highlighted the request to complete the survey in plenary sessions, and incentives (entry into a drawing to win free CME course tuition or restaurant gift certificates) were offered. Completion of the survey was voluntary and anonymous for all participants. This project was approved by the institutional review board of Massachusetts General Hospital.

Survey Instrument

The survey instrument addressed 4 domains of interest: (1) general knowledge of medical genetics, (2) knowledge of psychiatric genetics, (3) current practice patterns, and (4) attitudes toward clinical and ethical dilemmas that may arise in the application of genetic knowledge in psychiatry. A final section gathered demographic information. The survey contained a total of 26 knowledge, attitude, and practice pattern questions. All questions were written in multiple choice (single question and multiple parts), true/false, or fill-in-the-blank formats. A copy of the complete survey is available from the corresponding author (C.T.F.) upon request.

Scale Development

The survey was developed by the generation and selection of face-valid questions for each of the 4 domains. Experts contributing to the development and selection of questions included medical geneticists, clinical psychiatrists, and investigators in the field of psychiatric genetics. The survey was distributed to an initial pilot sample of approximately 30 psychiatrists and geneticists who were asked to comment on the readability, comprehensibility, and format of the survey. A second pilot sample of 241 psychiatrists, genetic counselors, genetic nurses, and geneticists were also administered the survey. Responses gathered from pilot samples of psychiatrists were not used in the final data analysis, although the knowledge scores of genetic counselors are presented in the discussion section for comparison. Selection and refinement of questions occurred following both phases of pilot testing. For the general genetic knowledge section, 9 questions were chosen to reflect major topics of Mendelian genetics, inheritance patterns, risk assessment, interpretation of test results, and understanding of statistics used in current genetics literature. The 12 items included in the psychiatric genetic knowledge section covered topics such as complex genetics mechanisms, estimation of recurrence risks (risk of developing a psychiatric condition among firstdegree relatives of patients with a given disorder), relative genetic and environmental contributions to psychiatric

illness, and genetic conditions that may present with psychiatric symptoms (e.g., velocardiofacial syndrome). In the attitudes and opinions section, 4 questions asked about uses of genetic testing in psychiatry, genetic counseling recommendations in a prenatal setting, and risks and benefits associated with increased genetic knowledge in psychiatry. The final 6 questions addressed current practice patterns and incorporation of genetic knowledge into clinical practice.

Analysis

Standard descriptive univariate and bivariate statistical methods were used to describe the data. Univariate correlations were computed using Spearman rank correlations. Nonparametric (Kruskal-Wallis and Mann-Whitney ranksum) tests were used to compare groups on the summary scores obtained. Logistic regression was used to examine the relationship between genetic knowledge and nondirectiveness in a hypothetical genetic counseling scenario. Although the focus of this article is on the responses of psychiatrists, we also compared general genetic knowledge scores to those of 2 samples that had been administered the same set of general genetic knowledge items: (1) the pilot sample of genetic counselors (N = 149) noted above and (2) a sample of endocrinologists (N = 601) who were administered the same items as part of another study in which they completed an online survey prior to attending a national meeting of endocrinologists (M. Freeman, M.D.; W. Crowley, M.D.; V. Hughes, M.S.; et al., unpublished data, 2002).

RESULTS

A total of 352 psychiatrists (54% of those surveyed) returned completed or partially completed surveys. Answers that were inconsistent (e.g., multiple responses to single-response questions) were excluded. All remaining available answers were used in the analysis, and the number of available answers varied for each question.

Demographics

All respondents were psychiatrists (97%) or in psychiatric training (3%) at the time of the completion of the survey. Table 1 summarizes the demographic characteristics of the sample. As shown, more than half of the sample (53%) had completed psychiatric residency training greater than 15 years prior to the survey. Respondents included psychiatrists working in a broad range of health care positions and clinical settings.

Psychiatric Genetic Knowledge

The median number of questions answered correctly in this section was 4 (33.3%), with a range of 0 to 10. No respondent answered all questions correctly, and only 15.5% of psychiatrists answered at least half of the ques-

Table 1. Demographic Characteristics of 352 Psychiatrist	
Survey Respondents	

Characteristic	Sample, %
Sex	
Men	58.7
Women	41.3
Board certified in psychiatry	85.4
Years since completion of residency training ^a	
Not yet completed	2.8
0-5	11.7
6–10	15.1
10–15	17.9
16–20	14.2
> 20	38.4
Major work activity	
Hospital or inpatient psychiatry	20.9
Outpatient psychopharmacology	51.7
Psychotherapy	14.6
Research	0.3
Administration	3.6
Other	8.9
Setting of clinical practice	
Department of psychiatry in general hospital	17.8
Public psychiatric hospital	7.5
Private psychiatric hospital	4.6
Community clinic/mental health center	15.6
Home or private office	34.9
Multispecialty group practice	6.2
None	0.9
Other	12.5
^a The ranges reflect the original wording of the sur	vey.

tions correctly. In a more conservative examination of responses, the scores were reanalyzed after eliminating those psychiatrists who did not answer all questions in this section; the median score among those who completed all items remained low at 5/12 (42%). Among the questions that psychiatrists scored poorly on were those asking about the role of specific genes in the development of Alzheimer's disease; only 6% to 14% of participants responded correctly to questions about genes known to be involved in the early- (presenilin-1, presenilin-2, amyloid precursor protein) and late-onset (apolipoprotein E) forms of the disease. Knowledge scores were inversely related to years since finishing training ($r_s = -0.166$, p = .002). Scores on the psychiatric genetic knowledge section were also slightly higher among men than women (median score = 5 vs. 4, respectively, p = .023), but did not differ by board certification status, setting of clinical practice, or principal activity at work.

One set of questions in the psychiatric genetics knowledge section with direct relevance to genetic counseling asked about the offspring recurrence risks (i.e., risk to an offspring of an affected parent) of 4 major psychiatric disorders: schizophrenia, bipolar disorder, major depression, and panic disorder. Although respondents gave a wide range of estimates, median responses were close to widely cited empiric estimates available in the medical literature (Table 2).^{1,23–28} The psychiatric genetics knowledge section also included a question asking respondents to estimate the proportion of disorder risk attributable to

Table 2. Estimation of Offspring Recurrence Risk by 352 Psychiatrist Survey Respondents

A woman is affected with a psychiatric illness and is currently pregnant with her first child. Her husband is unaffected. What is the approximate percent probability (0-100) that her child will be affected with the same disorder if she has:

Disorder	Median Response, %	Range of Responses, %	Estimated Recurrence Risk Derived From Medical Literature for First-Degree Relatives, %	Reference No.
Schizophrenia	13	1-80	5-16	1,23,24
Bipolar disorder	20	0.8-80	4-18	25
Major depression	25	1-80	5-25	1,26
Panic disorder	20	0–95	8-17	27,28

Table 3. Estimation of Heritability for Psychiatric and Medical Conditions by 352 Psychiatrist Survey Respondents

In the general population, what percentage of risk of developing the following conditions is due to heritable genetic factors?

Disorder	Median Response, %	Range of Responses, %	Estimates Derived From Medical Literature, %	Reference No.
Schizophrenia	30	0.2–100	70–86	24
Bipolar disorder	40	0.5-100	59-80	1,25
Major depression	30	1-100	21-45	1
Autism	10	0-100	> 90	1,29
Alzheimer's disease	20	0-100	60 (late onset)	30
Attention-deficit/ hyperactivity disorder	25	0–100	75–98	1
Alcoholism	30	2-90	47-75	31
Panic disorder	25	0-100	35-46	1,28
Antisocial personality disorder (in males)	20	0-100	41–69	32,33
Breast cancer	25	0.5-100	27	34

heritable genetic factors for a variety of psychiatric or medical conditions. (This question was not included in the generation of a psychiatric genetic knowledge score due to the imprecision and variation of estimates reported in the medical literature.) Table 3 summarizes the results and provides a comparison to heritability estimates obtained from widely cited published data.^{1,24,25,29-34} Respondents' estimates of heritability of psychiatric disorders were less consistent with published data compared to their responses regarding recurrence risks. Overall, respondents tended to underestimate the genetic contribution to major psychiatric illness, especially for schizophrenia, autism, Alzheimer's disease, and attention-deficit/hyperactivity disorder (ADHD). Interestingly, the disorder most often cited as having no genetic contribution was autism (7.7% of respondents); in fact, autism is the most highly heritable of the disorders listed. Also of interest, autism was also the disorder for which the greatest proportion of psychiatrists estimated 100% heritability (2.0% of respondents).

General Genetic Knowledge

The median number of questions answered correctly in this section was 4 (44.4%), with a range of 0 to 9. Eighteen percent of psychiatrists answered all 9 questions incorrectly, and fewer than 1% answered all questions correctly. Eliminating psychiatrists who missed questions in this section did not alter the median knowledge score. There was an inverse relationship between general knowledge scores and years since completion of residency training ($r_s = -0.206$, p < .001). General genetic knowledge scores were not significantly associated with sex, board certification status, setting of clinical practice, or principal activity at work.

Attitudes and Opinions Regarding Genetics in Psychiatry

Participants were asked questions regarding the risks, benefits, and use of genetic information in psychiatry. The majority of psychiatrists felt that there were both risks and benefits associated with increased knowledge about the genetic basis of psychiatric illness (Table 4). Improved understanding of the biological basis of disease and the potential for development of new and improved treatments were the most commonly cited benefits (chosen by 91% and 79% of psychiatrists, respectively). Denial of insurance (93% of respondents) and potential for employment discrimination (78% of respondents) were the most commonly cited risks. To examine how psychiatrists might make use of genetic information in clinical practice, respondents were asked how they would counsel a couple presenting for prenatal testing for psychiatric and medical disorders. Overall, 25% to 33% of psychiatrists indicated that they would give directive recommendations

Table 4. Perceived Risks and Benefits Associated With Genetic Knowledge in Psychiatry According to 352 Psychiatrist Survey Respondents

Which of the following, if any, do you think are likely to be BENEFITS of increased knowled (Please choose all that apply.)	ge of the genetic basis of psychiatric illness?
Response	% Choosing Response
Improved understanding of the biological basis of mental illness	91
Discovering new and better treatments	79
Diagnostic clarification of affected patients	77
Predictive testing for presymptomatic patients at risk	77
Targeting of resources to at-risk populations	74
Prenatal testing to guide reproductive choices	66
Destigmatization of mental illness	58
Elimination of mental illness	12
There are no benefits	< 1
Other	3
Which of the following, if any, do you think are likely to be RISKS of increased knowledge of	f the genetic basis of psychiatric illness?

(Please choose all that apply.) % Choosing Response Response Denial of insurance to patients with high-risk genes 93 Employment discrimination against people who carry genes for psychiatric illness 78 Lowered expectations for children who carry high-risk genes 63 52 Movement of research and clinical resources away from psychosocial therapies 36 Denial of resources to symptomatic patients who do not carry high-risk genes 35 Increased stigmatization of mental illness 33 Elimination of desirable traits through genetic selection against genes that contribute to mental illness 14 Elimination of mental illness through genetic selection against genes that contribute to mental illness 2 There are no risks 2 Other

Table 5. Genetic Counseling Recommendations by 352 Psychiatrist Respondents

You are meeting with a couple who requested prenatal testing due to a family history of a psychiatric or medical disorder. The test has revealed that the fetus will develop the disorder with high probability. When discussing the test results with the couple, would you advise termination of pregnancy, continuation of pregnancy, or provide nondirective information for each of the following disorders?

	Continuation,	Termination,	Nondirective
Disorder	%	%	Information, %
Schizophrenia	16	12	72
Bipolar disorder	22	3	75
Major depression	28	2	71
Autism	9	20	71
Alzheimer's disease	23	3	74
Attention-deficit/	29	1	69
hyperactivity disorder			
Alcoholism	27	2	71
Panic disorder	33	< 1	67
Antisocial personality	19	10	71
disorder			
Anencephaly	3	63	34
Huntington's disease	6	25	69
Down syndrome	7	19	74
Obesity	30	< 1	70

(recommending termination or continuation of pregnancy) in a hypothetical counseling scenario in which prenatal testing indicated a high likelihood of developing a psychiatric disorder (Table 5). Psychiatric disorders for which termination of pregnancy was most often recommended were autism (20%), schizophrenia (12%), and antisocial personality disorder (10%). To examine whether degree of genetic knowledge was associated with the tendency to be nondirective, a composite genetic knowledge score representing the sum of the general and psychiatric genetic knowledge scores was calculated for 4 major psychiatric disorders. Using univariate logistic regression,we found that higher scores on genetic knowledge were significantly associated with nondirectiveness in a hypothetical counseling scenario for all 4 disorders: schizophrenia (p < .001), bipolar disorder (p = .003), major depression (p = .002), and autism (p = .034).

Participants were also asked under what circumstances genetic testing would be appropriate for schizophrenia, bipolar disorder, and panic disorder, assuming a highly predictive test (Table 6). Decisions regarding the appropriateness of testing in those settings varied significantly depending on disorder, with testing being favored most often for schizophrenia and bipolar disorder in all scenarios. Between 2% and 10% of psychiatrists thought that genetic tests should never be used for any of the 3 disorders. Respondents were significantly more likely to favor prenatal testing in affected families in comparison to the general population for all 3 disorders (p < .001). Opinions regarding testing of adults and children did not differ significantly for predictive testing of asymptomatic individuals or clarification of diagnoses in affected individuals.

Practice Patterns

Psychiatrists were asked questions regarding the role of genetics in their clinical practice and their utilization of genetics professionals for consultation. Eighty-seven per-

Table 6	Hypothetical	Use of Genetic	· Testing Accord	ing to 352 Pe	wchiatrist Survey	Respondents
Table 0.	riypoinciicai	Use of deficite	. Itsting Accord	ing to 554 1 a	sycillati ist Surve	y nesponaents

If a genetic test with a high predictive probability (>95% chance of having a disorder with a positive test result) were available for each of the following disorders, in which clinical situations should the test be used?

Schizophrenia, %	Bipolar Disorder, %	Panic Disorder, %
70	58	35
19	14	10
45	43	29
47	44	30
75	74	66
75	72	63
	Schizophrenia, % 70 19 45 47 75 75	Schizophrenia, % Bipolar Disorder, % 70 58 19 14 45 43 47 44 75 74 75 72

cent indicated that they routinely take detailed family histories of psychiatric illness as part of their evaluation of patients. In addition, most psychiatrists reported discussing a genetic or hereditary component of psychiatric illness with some of their patients (Table 7).

The majority of psychiatrists (71%) had not referred any patients to a geneticist or genetic counselor for consultation in the past year, and an additional 20% had referred only 1 to 2 patients (Table 7). When presented with a variety of hypothetical clinical scenarios, psychiatrists showed varying tendencies to refer patients to genetic counselors (Table 7). Respondents were most likely to refer mentally ill patients with associated multiple congenital anomalies (94%) or mental retardation (82%).

Eighty-three percent of psychiatrists indicated that they feel it is their role to discuss genetic information about psychiatric illness with patients and families (Table 7). Surprisingly, however, a minority of psychiatrists reported feeling competent (23%) or adequately trained (15%) to do so (Table 7). Psychiatrists who reported feeling competent to discuss genetic information did score significantly higher on measures of general genetic knowledge (p = .012) and psychiatric genetic knowledge (p = .006) compared to those who did not feel competent. However, there were no differences in knowledge scores between respondents who agreed that their medical training had prepared them to discuss genetic information with patients compared to those who disagreed. This may be due in part to the relatively small number (N = 46) of respondents who felt that their training had prepared them for this role. There was no relationship between years since completion of residency training and feeling adequately trained ($r_s = 0.06$, p = .33) or competent ($r_s =$ 0.07, p = .25) to discuss genetics topics with patients.

The survey asked about exposure to sources of information about genetics. Most psychiatrists (40%) reported reading the current medical literature as their primary source of genetic information. Other sources included classes taken in medical school or in clinical training (27%), CME courses (22%), informal consultation with colleagues (7%), popular press or media (1%), or other (3%). When asked about educational opportunities, most psychiatrists expressed interest in all of the options presented, including written educational materials (93%), CME courses (82%), Web/Internet resources (81%), and multidisciplinary consultation with colleagues (77%).

DISCUSSION

To our knowledge, this study represents the largest survey of psychiatrists' knowledge of genetics and their attitudes toward the use of genetic information in clinical practice. The response rate was comparable to or exceeded that in other studies of physician attitudes and/or knowledge of genetics.^{13,35–37} With the accelerating pace of genetic research in neuropsychiatry, the coming decade will likely see increasing demand for the translation of genetic research into the clinical arena. An understanding of the utility and limitations of genetic research for guiding risk estimation, diagnostic clarification, and treatment planning will become an important part of clinicians' skill sets. The results of our survey therefore bear on important issues related to psychiatrists' preparation for the impact of genetics on clinical psychiatry. Several findings are of particular note. First, most psychiatrists responding to this survey evidenced gaps in their knowledge of genetic principles and psychiatric genetic findings. Second, most believe it is their role to discuss familial and genetic aspects of psychiatric illness with patients and families but feel unprepared to do so. Third, these psychiatrists appear to make limited use of referral to genetics professionals. Fourth, a subset of respondents expressed attitudes about potential uses of genetic information that vary from standards endorsed by genetics professionals. Fifth, most psychiatrists expressed interest in further education in genetics. As we discuss below, each of these findings has implications for how the field of psychiatry may respond as increased insight into the genetic determinants of psychiatric disorders is gained.

Psychiatrists' general knowledge of genetics was assessed with a series of questions covering several topics including Mendelian inheritance patterns, risk estimation, principles of interpreting genetic test results, and the general meaning of statistical measures commonly reported in genetic studies. We and others³⁸ consider these topics to be basic concepts central to the application of genetics in clinical practice. The assessment of psychiat-

Table 7.	Practice	Patterns	of 352	Psychiatrist	Survey
Respon	dents				

How often, if ever, do you discuss with patients the extent to which there is a genetic or hereditary component to the patient's psychiatric illness?

Response	% Choosing Response
With all of my patients	10
With most of my patients	25
With some of my patients	58
With none of my patients	5
Not applicable no clinical practice	2

Over the past year, how many psychiatric patients have you referred for consultation regarding a genetic component of psychiatric illness in themselves or family members?

Response	% Choosing Response
0	71
1–2	20
3–4	4
5-10	3
> 10	1
Not applicable	2

For the following clinical situations, indicate whether you would address the issue yourself or refer the patient to a genetic counselor if available.

		Refer to
	Address	Genetic
Situation	Myself, %	Counselor, %
A patient wonders about the risk of	62	38
passing on mental illness to her child		
A couple with a family history of	33	67
mental illness who has		
family-planning questions		
A patient with mental illness and multiple congenital anomalies	6	94
A patient with mental illness and mental retardation	18	82
A family with multiple members affected with the same mental illness	42	58
A woman with questions about the safety	88	12
of medication use during pregnancy		
Statement	Agr	ee, %
I feel competent to discuss genetic	2	23
information regarding psychiatric illness with patients and their families		
I feel it is my role to discuss genetic	:	83
information regarding psychiatric illness		
My medical training has prepared me to		15
discuss genetic information regarding		15
nevchiatric illness with patients and		
their families		

ric genetic knowledge covered widely available information about the familial and genetic aspects of neuropsychiatric disorders. Overall, respondents appeared to have gaps in knowledge in these areas, with a median score of 44% on the general genetic knowledge items and 33% on the psychiatric genetic knowledge items. These findings are consistent with previous evidence about the degree to which psychiatrists are informed about genetics. In a survey of genetic knowledge among primary care physicians (internists, pediatricians, family practice physicians, and obstetrician-gynecologists) and psychiatrists, Hofman and colleagues¹⁴ reported that although all physicians scored significantly lower on genetic knowledge than a comparison group of genetics professionals, psychiatrists scored the lowest of all groups. Although our survey (completed 11 years later) is not directly comparable, the continued evidence of knowledge gaps highlights the need for education about genetics among psychiatrists.

Debates about the importance of "nature versus nurture" in human behavior and mental illness have had a long and sometimes contentious history in both professional circles and popular culture. Our results suggest that substantial differences of opinion remain among psychiatrists in that estimates of the heritability of psychiatric illnesses varied widely in our sample (Table 3). For the most part, however, psychiatrists tended to underestimate the contribution of genetic factors, as evidenced by the fact that median heritability estimates were generally lower than those derived from available twin studies.

In our survey, several of the items in the psychiatric genetic knowledge section dealt with issues that may be directly relevant to clinical practice. For example, knowledge of recurrence risks for relatives of individuals affected with psychiatric disorders is currently the most useful basis for prediction of illness risk and genetic counseling²²; although median responses were in line with empirical risks, respondents' estimates of recurrence risks varied widely for 4 common psychiatric disorders. Misconceptions regarding familial risk of psychiatric illness demonstrate 1 area where clinicians may be unaware of clinically relevant information. Another area with relevance to current clinical practice is the genetic basis of Alzheimer's disease. Four specific genes are known to play a role in the etiology of early- or late-onset Alzheimer's disease, and genetic testing is currently commercially available in the United States for 1 of them (presenilin 1, associated with early-onset autosomal dominant Alzheimer's disease). Among psychiatrists responding to this survey, a minority (< 15%) were able to identify the mode of inheritance of the 4 known Alzheimer's disease genes. Psychiatrists' limited knowledge in this area may lead to missed opportunities for referral of affected or at-risk patients for appropriate testing and genetic consultation.

Perhaps the most striking of our results was the finding that more than 80% of psychiatrists reported that discussing genetic information with patients and families is part of their professional role, but fewer than 25% felt prepared or competent to do so. This "preparedness" problem is likely not unique to psychiatry. Surveys involving internists and primary care physicians have also suggested that familiarity with genetics among these physicians is limited.^{12–14,36,37,39,40} In a recent survey of chairpersons of departments of medicine, only 25% believed that internists in their departments had enough general knowledge about genetic conditions to provide appropriate genetic counseling.⁴⁰ Another survey found that only 29% of physicians

felt they were qualified to provide genetic counseling regarding cancer risk to their patients.⁴¹ With the rapid acceleration of genetic research and the promise of "genomic medicine," numerous commentators have argued that there is a critical need for education of clinicians about evaluating and communicating familial and genetic information.^{22,42,43} Our results document an unmet need for education about genetics among practicing psychiatrists.

We also found evidence that psychiatrists rarely seek consultation from genetics professionals. More than 90% of respondents said that they had referred 2 or fewer patients for genetics consultation over the past year. On the other hand, for several clinical situations, most indicated that they would refer patients to a genetic counselor rather than address the issue themselves. It is unclear whether this disparity is due to psychiatrists not encountering these situations in clinical practice, not having access to genetic services, or not knowing how to access available consultation services. Nevertheless, the willingness of psychiatrists to refer to genetics professionals highlights a potential demand for these services.

Given that most of our respondents considered it their role to discuss genetic information with patients and families and rarely referred patients for genetic consultation, their responses to questions about hypothetical genetic counseling and genetic testing scenarios may give an indication of how psychiatrists would incorporate genetics into clinical practice. In response to questions regarding counseling a couple about genetic tests for psychiatric illnesses, approximately 30% of the sample said they would offer directive advice about continuing or terminating a pregnancy. A nondirective approach has been the cornerstone of traditional genetic counseling,^{15–18} but our results are consistent with other studies suggesting that nongenetics specialists may be more likely to be directive than are genetics professionals.¹⁹⁻²¹ Of note, in our survey, psychiatrists who scored higher on genetic knowledge were more likely to be nondirective in their responses to a hypothetical counseling scenario. This suggests that education about genetics may result in a greater appreciation of the manner in which genetic information is appropriately conveyed.

In our survey, a substantial proportion of psychiatrists felt that—given a highly predictive test—genetic testing for schizophrenia, bipolar disorder, and to a lesser extent, panic disorder would be appropriate for diagnostic clarification in symptomatic adults and children. In addition, predictive testing of asymptomatic individuals with a family history of these disorders was also considered useful by 29% to 47% of respondents. It is noteworthy that the willingness to test did not appear to depend on whether the subject was an adult or a child. Geneticists and medical ethicists have traditionally been cautious about the use of genetic testing in minors unless there is a clear medical benefit. Although earlier detection and treatment of psychiatric illness may influence clinical course,^{44–47} the ethical, legal, and psychosocial implications of genetic testing for minors are complex and require careful consideration.^{48–53}

Psychiatrists also indicated interest in the use of tests for prenatal screening in affected patients (35%-70%) and the general population (10%-19%) depending on the disease in question. A previous study that included a small sample of U.K. psychiatrists (N = 32) indicated that psychiatrists may have less favorable attitudes toward prenatal testing for bipolar disorder than do patients affected with the illness.⁵⁴ On the other hand, at least 1 previous study has documented that psychiatrists are more likely to routinely favor genetic testing than are other primary care physicians. Faden et al.55 surveyed 1140 physicians regarding their views on screening for cystic fibrosis using a test that identified only 80% of carriers. Among respondents, psychiatrists were the only group in which the majority favored routine testing. The authors conclude that lack of familiarity with cystic fibrosis and genetic testing (psychiatrists had the lowest mean knowledge scores) may have contributed to increased use of testing.55 An additional interpretation is supported by a secondary analysis undertaken by the same group, which showed that tolerance for ambiguity was higher among psychiatrists in comparison to other medical specialties.⁵⁶ Although lack of knowledge of appropriate uses of testing cannot be overlooked, psychiatrists, who are currently without any confirmatory diagnostic testing to support clinical practice, may have different perceptions of the value of a test with an 80% detection rate in comparison to other medical specialists.

Finally, our survey asked about psychiatrists' exposure to genetic information and interest in further genetics education. The most common source of information about genetics reported by respondents was reading the medical literature. Given the findings of our survey, current sources appear to be inadequate. The overwhelming majority of respondents (77%–93%) indicated interest in a variety of opportunities for education including CME courses, written and Web-based materials, and multidisciplinary consultation with colleagues. Active efforts should be directed toward the development of educational programs for psychiatrists and other nongeneticist physicians to maximize opportunities for the translation of research findings regarding the genetic basis of illness into patient care.

This study had several limitations. First, the sample comprised psychiatrists attending a CME course, and therefore may have ascertained a group of psychiatrists more likely to take an active interest in ongoing education. Additionally, the content of the CME course (an update on psychopharmacology) may also have selected a population of psychiatrists who are not representative of practicing psychiatrists in general. Furthermore, psychia-

trists were presented with a variety of hypothetical clinical situations in which use and interpretation of genetics tests with high predictive probability were examined. Currently, no such testing exists (and it may never exist) for psychiatric disorders, and psychiatrists would not have encountered similar situations in training or clinical practice. Susceptibility genes for psychiatric disorders are expected to have modest individual effects, and professional groups have consistently recommended against providing clinical testing for genes (such as the apolipoprotein E4 polymorphism in Alzheimer's disease) that have low positive predictive value for neuropsychiatric disorders.⁵⁷ On the other hand, there may be subtypes of psychiatric illness (e.g., psychotic disorders associated with velocardiofacial syndrome) for which available tests may provide useful diagnostic clarification.⁵⁸ Moreover, recent analyses suggest that combining data from multiple susceptibility loci may provide substantial positive predictive value for complex disorders.59

Second, it is possible that respondents differed from nonrespondents in their knowledge and attitudes about genetics. Unfortunately, data on nonrespondents were not available to address this question. However, as we have noted, our response rate was comparable to or greater than those of other physician surveys regarding genetics. In addition, because the study lacked a formal comparison group, it is unknown how genetics professionals would have responded to the same or similar questions. It might be argued that psychiatrists' low scores on genetic knowledge items were a result of particularly demanding or obscure questions. Some indication of the level of difficulty and relevance of the questions can be gathered from analysis of data obtained from the survey of genetic counselors during the pilot phase of the study. Although the response rate for this group was lower, and the pilot survey was longer, all genetic knowledge questions in the final version of this survey were also answered by the genetic counselors. The genetic counselors (N = 149) scored significantly higher than the psychiatrists on psychiatric genetics knowledge (median correct: 66.7% vs. 33.3%, p < .0001) and general genetics knowledge (median correct: 77.8% vs. 44.4%, p < .0001), suggesting that the items captured information within the knowledge base of genetic counselors. The general genetics questions were also administered to a group of endocrinologists (N = 601) attending the 2002 85th annual meeting of the Endocrine Society (M. Freeman, M.D.; W. Crowley, M.D.; V. Hughes, M.S.; et al., unpublished data, 2002). The median score among the endocrinologists was significantly higher than the psychiatrists' scores on the same set of questions (median correct: 55.6% vs. 44.4%, p < .0001).

This study was an initial attempt to determine the preparedness of practicing psychiatrists to begin to incorporate genetic information into the clinical care of their patients. With rapid advances in psychiatric genetics, there is likely to be a growing need for clinicians who can provide patients with up-to-date genetic counseling. The question of how and by whom this counseling is best provided remains open. While genetic counselors and medical geneticists may be most experienced with the interpretation of complex inheritance patterns and the nondirective interpretation of genetic risk information, psychiatrists are the clinicians most likely to first encounter patients, to take a detailed family history, and to assess the need for further evaluation or consultation. In addition, psychiatrists are best able to discuss the variable clinical course and treatment options of psychiatric illness with patients. Our study suggests that psychiatrists have a great need and correspondingly strong desire for further education in genetics.

REFERENCES

- Genetics and mental disorders. National Institute of Mental Health's Genetics Workgroup. Biol Psychiatry 1999;45:559–573
- McGuffin P, Owen MJ, Gottesman II. Psychiatric Genetics and Genomics. Oxford, England: Oxford University Press; 2002
- Blacker D, Tanzi RE. The genetics of Alzheimer disease: current status and future prospects. Arch Neurol 1998;55:294–296
- Harrison PJ, Owen MJ. Genes for schizophrenia? recent findings and their pathophysiological implications. Lancet 2003;361:417–419
- Smith LB, Sapers B, Reus VI, et al. Attitudes towards bipolar disorder and predictive genetic testing among patients and providers. J Med Genet 1996;33:544–549
- Milner KK, Collins EE, Connors GR, et al. Attitudes of young adults to prenatal screening and genetic correction for human attributes and psychiatric conditions. Am J Med Genet 1998;76:111–119
- Milner KK, Han T, Petty EM. Support for the availability of prenatal testing for neurological and psychiatric conditions in the psychiatric community. Genet Test 1999;3:279–286
- Trippitelli CL, Jamison KR, Folstein MF, et al. Pilot study on patients' and spouses' attitudes toward potential genetic testing for bipolar disorder. Am J Psychiatry 1998;155:899–904
- Turlington L, Moldin SO. Assessing the demand for psychiatric genetic counseling [abstract]. Am J Hum Genet 1994;A144
- Peay H, McInerney J. A pilot study on psychiatric genetic counseling: counselor's needs [abstract]. J Genet Couns 2002;11:485
- Menasha JD, Schechter C, Willner J. Genetic testing: a physician's perspective. Mt Sinai J Med 2000;67:144–151
- Firth HV, Lindenbaum RH. UK clinicians' knowledge of and attitudes to the prenatal diagnosis of single gene disorders. J Med Genet 1992;29: 20–23
- Wilkins-Haug L, Hill LD, Power ML, et al. Gynecologists' training, knowledge, and experiences in genetics: a survey. Obstet Gynecol 2000; 95:421–424
- Hofman KJ, Tambor ES, Chase GA, et al. Physicians' knowledge of genetics and genetic tests. Acad Med 1993;68:625–632
- Kessler S. Genetic counseling is directive? look again [letter]. Am J Hum Genet 1997;61:466–467
- Kessler S. Psychological aspects of genetic counseling, 11: nondirectiveness revisited. Am J Med Genet 1997;72:164–171
- Wertz DC, Fletcher JC. Attitudes of genetic counselors: a multinational survey. Am J Hum Genet 1988;42:592–600
- Juengst ET. Patterns of reasoning in medical genetics: an introduction. Theor Med 1989;10:101–105
- Carnevale A, Lisker R, Villa AR, et al. Counselling following diagnosis of a fetal abnormality: comparison of different clinical specialists in Mexico. Am J Med Genet 1997;69:23–28
- Marteau T, Drake H, Bobrow M. Counselling following diagnosis of a fetal abnormality: the differing approaches of obstetricians, clinical geneticists, and genetic nurses. J Med Genet 1994;31:864–867
- 21. Geller G, Tambor ES, Chase GA, et al. Incorporation of genetics in

primary care practice: will physicians do the counseling and will they be directive? Arch Fam Med 1993;2:1119–1125

- Merikangas KR, Risch N. Will the genomics revolution revolutionize psychiatry? Am J Psychiatry 2003;160:625–635
- Tsuang M. Schizophrenia: genes and environment. Biol Psychiatry 2000;47:210–220
- Tsuang MT, Stone WS, Faraone SV. Genes, environment and schizophrenia. Br J Psychiatry 2001;178(suppl 40):S18–S24
- Smoller JW, Finn CT. Family, twin and adoption studies of bipolar disorder. Am J Med Genet C Semin Med Genet 2003;123:48–58
- Sullivan PF, Neale MC, Kendler KS. Genetic epidemiology of major depression: review and meta-analysis. Am J Psychiatry 2000;157: 1552–1562
- Hettema JM, Neale MC, Kendler KS. A review and meta-analysis of the genetic epidemiology of anxiety disorders. Am J Psychiatry 2001;158:1568–1578
- Smoller JW, Finn CT, White CA. The genetics of anxiety disorders: an overview. Psychiatr Ann 2000;30:745–753
- Folstein SE, Rosen-Sheidley B. Genetics of autism: complex aetiology for a heterogeneous disorder. Nat Rev Genet 2001;2:943–955
- Bergem AL, Engedal K, Kringlen E. The role of heredity in late-onset Alzheimer disease and vascular dementia: a twin study. Arch Gen Psychiatry 1997;54:264–270
- Heath AC, Bucholz KK, Madden PA, et al. Genetic and environmental contributions to alcohol dependence risk in a national twin sample: consistency of findings in women and men. Psychol Med 1997;27: 1381–1396
- Fu Q, Heath AC, Bucholz KK, et al. Shared genetic risk of major depression, alcohol dependence, and marijuana dependence: contribution of antisocial personality disorder in men. Arch Gen Psychiatry 2002;59: 1125–1132
- Kendler KS, Prescott CA, Jacobson K, et al. The joint analysis of personal interview and family history diagnoses: evidence for validity of diagnosis and increased heritability estimates. Psychol Med 2002;32: 829–842
- 34. Lichtenstein P, Holm NV, Verkasalo PK, et al. Environmental and heritable factors in the causation of cancer: analyses of cohorts of twins from Sweden, Denmark, and Finland. N Engl J Med 2000;343:78–85
- Mountcastle-Shah E, Holtzman NA. Primary care physicians' perceptions of barriers to genetic testing and their willingness to participate in research. Am J Med Genet 2000;94:409–416
- Hunter A, Wright P, Cappelli M, et al. Physician knowledge and attitudes towards molecular genetic (DNA) testing of their patients. Clin Genet 1998;53:447–455
- Acton RT, Burst NM, Casebeer L, et al. Knowledge, attitudes, and behaviors of Alabama's primary care physicians regarding cancer genetics. Acad Med 2000;75:850–852
- Core Competencies in Genetics Essential for All Health-Care Professionals. Lutherville, Md: National Coalition for Health Professional Education in Genetics; 2001
- Escher M, Sappino AP. Primary care physicians' knowledge and attitudes towards genetic testing for breast-ovarian cancer predisposition. Ann Oncol 2000;11:1131–1135
- Taylor MR. A survey of chairpersons of departments of medicine about the current and future roles of clinical genetics in internal medicine. Genet Med 2003;5:328–331

- Freedman AN, Wideroff L, Olson L, et al. US physicians' attitudes toward genetic testing for cancer susceptibility. Am J Med Genet 2003; 120A:63–71
- Khoury MJ. Genetics and genomics in practice: the continuum from genetic disease to genetic information in health and disease. Genet Med 2003;5:261–268
- Guttmacher AE, Collins FS. Genomic medicine: a primer. N Engl J Med 2002;347:1512–1520
- 44. McGlashan TH, Zipursky RB, Perkins D, et al. The PRIME North America randomized double-blind clinical trial of olanzapine versus placebo in patients at risk of being prodromally symptomatic for psychosis, 1: study rationale and design. Schizophr Res 2003;61:7–18
- 45. Phillips LJ, Yung AR, Yuen HP, et al. Prediction and prevention of transition to psychosis in young people at incipient risk for schizophrenia. Am J Med Genet 2002;114:929–937
- Tsuang MT, Stone WS, Faraone SV. Understanding predisposition to schizophrenia: toward intervention and prevention. Can J Psychiatry 2002;47:518–526
- Cannon TD, Huttunen MO, Dahlstrom M, et al. Antipsychotic drug treatment in the prodromal phase of schizophrenia. Am J Psychiatry 2002;159:1230–1232
- Demmer LA, O'Neill MJ, Roberts AE, et al. Knowledge of ethical standards in genetic testing among medical students, residents, and practicing physicians [letter]. JAMA 2000;284:2595–2596
- Nelson RM, Botkjin JR, Kodish ED, et al. Ethical issues with genetic testing in pediatrics. Pediatrics 2001;107:1451–1455
- 50. Wertz DC, Fanos JH, Reilly PR. Genetic testing for children and adolescents: who decides? JAMA 1994;272:875–881
- 51. Points to consider: ethical, legal, and psychosocial implications of genetic testing in children and adolescents. American Society of Human Genetics Board of Directors, American College of Medical Genetics Board of Directors. Am J Hum Genet 1995;57:1233–1241
- Genetic testing for cystic fibrosis. National Institutes of Health Consensus Development Conference Statement on genetic testing for cystic fibrosis. Arch Intern Med 1999;159:1529–1539
- Mental Disorders and Genetics: The Ethical Context. Nuffield, England: Nuffield Council on Bioethics; 1998
- Jones I, Scourfield J, McCandless F, et al. Attitudes towards future testing for bipolar disorder susceptibility genes: a preliminary investigation. J Affect Disord 2002;71:189–193
- Faden RR, Tambor ES, Chase GA, et al. Attitudes of physicians and genetics professionals toward cystic fibrosis carrier screening. Am J Med Genet 1994;50:1–11
- Geller G, Tambor ES, Chase GA, et al. Measuring physicians' tolerance for ambiguity and its relationship to their reported practices regarding genetic testing. Med Care 1993;31:989–1001
- Relkin NR, Kwon YJ, Tsai J, et al. The National Institute on Aging/ Alzheimer's Association recommendations on the application of apolipoprotein E genotyping to Alzheimer's disease. Ann N Y Acad Sci 1996;802:149–176
- Hodgkinson KA, Murphy J, O'Neill S, et al. Genetic counselling for schizophrenia in the era of molecular genetics. Can J Psychiatry 2001; 46:123–130
- Yang Q, Khoury MJ, Botto L, et al. Improving the prediction of complex diseases by testing for multiple disease-susceptibility genes. Am J Hum Genet 2003;72:636–649