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Genetic services for common complex disorders: Surveys of health maintenance organizations and academic genetic centers

Melanie F. Myers, MS[1], Teresa Doksum, PhD, MPH[1,2], and Neil A. Holtzman, MD, MPH[1]

Purpose: To learn the extent to which HMOs and academic genetic centers (1) are involved in predictive genetic tests for common, complex disorders and (2) interact with each other. **Methods:** Surveys of HMO medical directors and directors of U.S. academic genetic centers. **Results:** In 1996, approximately 28% of HMOs were covering predictive tests for breast and colon cancer, but 75% of all medical directors said their HMO would consider policies regarding predictive testing in the next 5 years. Approximately 80% of directors of academic genetic centers said they provided genetic counseling services for common adult-onset disorders for patients covered by managed care organizations (MCOs), but they ranked the volume of services they provide for pediatric and prenatal indications much higher. Most academic genetic centers (72%) have contracts with MCOs. **Conclusion:** Although genetic services are being provided by academic genetic centers to patients who are members of managed care organizations, many patients with whom genetic testing for adult onset disorders is discussed may never see a geneticist. Academic genetic centers should educate nongeneticist professionals about the use of tests for common disorders. ***Genetics in Medicine*, 1999:1(6):272–285.**

Key Words: *Genetic services, health maintenance organizations, managed care organizations, predictive genetic testing, breast cancer, colon cancer, Alzheimer disease*

Within the past 10 years, tests for predicting genetic susceptibility to Alzheimer disease, breast cancer, colon cancer, and other common adult-onset disorders have been developed. The extent of demand for these tests should depend on the strength of the associations between genotypes and diseases (clinical validity) and evidence on the safety and effectiveness of interventions to reduce risk and/or improve outcomes (utility) in those found by testing to be at increased risk.[1] At present, clinical laboratories can market tests for which clinical validity and utility have not been established.[1]

Lack of data on validity and utility affects the willingness of health insurers, including managed care organizations, to cover predictive genetic tests.[2] In 1995, fewer than 16% of 166 health insurers had ever made a decision to cover breast/ovarian cancer (BRCA) susceptibility testing.[2] The high cost of these tests limits their diffusion into practice as many people cannot afford to pay for them out-of-pocket. Fear that insurers will discriminate against those with positive test results has also limited interest in testing,[3,4] although state and federal legislation has been slowly reducing the potential for discrimination.[5] Despite the limited diffusion, the rapid pace of genetic discoveries and increased media coverage suggest that predictive genetic testing could play an expanding role in medical care. We wanted to learn more about the state of such testing, as well as the involvement of geneticists, among health maintenance organizations (HMOs) and managed care organizations (MCOs).

In the first of the two surveys reported here, we attempted to learn the extent to which HMOs received requests for and were covering predictive genetic tests for breast cancer, colon cancer, and Alzheimer disease (AD), and whether they had established arrangements with genetic specialists. We selected HMOs because nearly 80 million people (over one-quarter of the population) were enrolled in them in the United States and Puerto Rico in 1996, and because their enrollment had increased 57.5% between 1993 and 1996.[6] HMOs were also more likely than preferred provider organizations, indemnity plans, or self-insured plans to cover established genetic technologies and other genetic services (Schoonmaker et al., unpublished data, 1999). At the time of the survey, tests for inherited susceptibility were available for mutations at the BRCA1 and 2 loci, at several different loci for hereditary nonpolyposis colon cancer, and for the apolipoprotein E-ϵ4 polymorphism, which had been associated with AD. However, the American Society of Human Genetics recommended that predictive testing for breast cancer should be conducted on an investigational basis[6] and several organizations opposed use of the ApoE-ϵ4 test to

From [1]Genetics and Public Policy Studies, The Johns Hopkins Medical Institutions, Baltimore, Maryland and [2]Abt Associates Inc., Cambridge Massachusetts.

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Neil A. Holtzman, MD, Genetics and Public Policy Studies, The Johns Hopkins Medical Institutions, 550 N. Broadway, Suite 511, Baltimore, MD 21205-2004.

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predict risk of Alzheimer disease as there was no treatment or prevention for the disease.[8]

We were also interested in learning the extent to which geneticists were providing services for adult-onset conditions. The existence of tests that detect inherited predispositions does not necessarily mean that geneticists will be the ones who offer or interpret them. The three common disorders for which tests are available are often managed by nongeneticist specialists in conjunction with primary care providers. These nongeneticists could offer predictive tests, as well as interpret the results. In order to learn more about geneticists' role we also surveyed directors of academic genetic units in the United States. Compared with private genetic units, academic genetic units are more readily identifiable and are also likely to be the genetic centers receiving the largest number of referrals.

We also used the two surveys to learn more about the relations between managed care organizations and clinical geneticists, and to learn in which laboratories genetic tests are being performed.

METHODS

Survey of HMOs

Written survey

A one-page questionnaire was mailed in September, 1996 to 535 medical directors listed in the directory of the American Association of Health Plans (AAHP).[9] Medical directors of HMOs who failed to respond were remailed the questionnaire. In the cover letter accompanying the survey, medical directors were asked to forward the questionnaire to someone else in their organization if that person was better able to complete it.

A preliminary draft of the questionnaire was pilot-tested in two stages. In the first, the HMO Group, a national association of nonprofit HMOs covering over six million HMO subscribers, agreed to fax a draft to its 29 member organizations. Based on the responses from 22 medical directors, the questionnaire was modified. The modified draft was sent to experts in the field of managed care. Their few suggestions were incorporated. Although wording differed, the pilot and final questionnaire were quite similar in content. Hence, the 22 HMO Group respondents were included in the analysis.

The final one-page questionnaire consisted of three sections of close-ended questions (See Appendix for the final questionnaire). The first section elicited information regarding four adult-onset disorders: breast/ovarian cancer, colon cancer, Alzheimer disease, and Huntington disease. For each disorder, we asked respondents to indicate (a) if they had ever received requests from providers for predictive genetic testing; (b) if they had ever covered the request; and c) if they had a written policy regarding coverage for predictive genetic testing. Although it is much less common, we included Huntington disease because it is an adult-onset disorder for which testing criteria are well-established. The second section asked respondents if they expected their HMO to consider developing policies regarding predictive genetic testing for adult disorders in the next 1–5 years. The third section asked respondents about the types of arrangements the HMO had established with genetic specialists (employ, contract with, or consult informally). Genetic specialists were defined as medical geneticists, genetic counselors, and nurses trained in genetics.

Telephone follow-up

Respondents to the one-page questionnaire who indicated that they either had covered predictive genetic testing for breast/ovarian cancer, colon cancer, or Alzheimer's disease (n = 42) or had coverage policies for predictive genetic testing (n = 8 HMO Group members) were contacted by telephone. As part of the phone interview, we asked respondents to confirm and/or clarify their responses to the written survey regarding requests for and coverage of predictive tests. We also asked about factors that were or would be important in establishing relationships with genetic specialists, as well as whether the HMO had developed any educational programs about genetics for nongenetic health care professionals or patients.

Classification of HMOs

Because of similarities between Network and Individual Practice Association (IPA) HMOs we merged these two categories for purposes of analysis. For the same reason, we also merged Group with Staff HMOs. Respondents that had enrollees in both of these two merged categories are referred to as "Mixed HMOs."

Survey of Directors of Academic Genetic Center Directors

Of the 105 programs listed in The Guide to North American Graduate and Postgraduate Training Programs in Human Genetics,[10] 69 in the U.S. listed "clinical genetics" as one of their areas of concentration. They were mailed questionnaires. A draft of the questionnaire was sent to the head of an academic genetics unit for "pilot" testing. His comments were incorporated into the final questionnaire. Some questions used wording parallel to those in the previously pilot-tested HMO questionnaire, and we did not want to change them.

The academic genetic center directors were asked close-ended or multiple choice questions, including whether they had contractual arrangements with MCOs, the types of prenatal, pediatric, and adult genetic services they provided to MCOs, the frequency of referrals from MCOs for these services, and issues of coverage they encountered in providing genetic services to patients in MCOs. We used "MCOs" rather than "HMOs" as the more encompassing term that did not require respondents to distinguish between types. The genetic center directors were also asked to indicate the source of patient referrals to them and if their unit had developed any educational materials or programs for nongenetic health care professionals or patients. (See Appendix for the final questionnaire).

RESULTS

SURVEY OF HMOS

Response rates

Including the HMO Group, responses were received from 211 of the 563 HMOs to whom the questionnaire was mailed (37%). The overall response rate for the phone interview was 78% (39/50). Based on the way they were classified in the AAHP database, 132 Network/IPA HMOs (35%), 19 Group/Staff HMOs (36%), and 34 Mixed HMOs (50%) responded. According to data in the AAHP database, respondent and non-respondent HMOs did not differ by the number of primary, specialty or total physicians with whom they contract, or whether they met federal qualifications, National Committee on Quality Assurance accreditation, or had a point of service plan.

Coverage of requests

Medical directors of 43% of HMOs reported receiving requests for predictive genetic testing for breast cancer compared with 26%, 23%, and 8% respectively for Huntington disease colon cancer and Alzheimer disease (Table 1). However, they were more likely to cover testing for Huntington Disease than breast cancer (Chi squared, $P = 0.025$), colon cancer ($P = 0.019$) or Alzheimer disease ($P < 0.001$). Only one HMO covered the test for Alzheimer disease. Network/IPA HMOs reported covering requests for predictive genetic tests for all three diseases more often than Group/Staff and Mixed HMOs (Fig. 1) but only for breast/ovarian cancer was the difference significant ($P < 0.001$).

Differences in mailed versus phone

We were surprised by the relatively large percentage of HMOs reporting that they had actually covered predictive genetic testing for BRCA and colon cancer in view of a 1995 survey showing very sparse coverage for BRCA testing.[2] Therefore, in the phone interview, we defined predictive genetic testing as testing conducted in healthy individuals to predict risk of future disease. After being given this definition, 12 of the 35 respondents changed their answers. In three of these cases, the person who completed the phone interview was different from the person who completed the written questionnaire. The change in response was uniformly in the direction of fewer requests and fewer decisions to cover. Table 2 summarizes this information and shows that the number of reported requests covered for predictive genetic testing for breast cancer fell from 29 in the written questionnaire to 21 in the phone survey, and those for colon cancer fell from 17 to 11.

Table 1
Coverage of requests for predictive genetic testing by HMOs

	Received requests		Covered*	
	n	%	n	%
Huntington disease	54	26	35	65
Breast cancer	88	43	40	45
Colon cancer	48	23	20	42
Alzheimer disease	17	8	1	6

*Among those that received requests.
n = 211.

Percent considering future polices by HMO type

In the written survey, 44% of HMOs reported that they would consider policies for predictive genetic testing in the next one year, and an additional 31% reported that they would consider policies in the next 2–5 years.

Arrangements with genetic specialists by model type

Most HMOs had established relationships with genetic specialists. The most frequently reported arrangement for all types of HMOs was to contract with genetic specialists (65%). We did not determine if these contracts reimburse specialists on a fee-for-service, salary, or capitation basis. Fewer HMOs reported informal arrangements with genetic specialists (35%) and only 6% reported employing genetic specialists. When examined by HMO type, Group/Staff reported more informal arrangements and more employment of genetic specialists than Network/IPA and Mixed HMOs (Fig. 2).

During the follow-up phone interview, respondents were asked to elaborate on the types of informal arrangements they had with genetic specialists. Most often respondents indicated that they either referred to genetic specialists or bought their services as needed. In a few cases, geography limited establishing formal arrangements with genetic specialists, as there were no genetic specialists available near the HMO.

Factors influencing HMOs to establish arrangements with specialists

During the phone interview, respondents who reported having any type of arrangement with genetic specialists were presented with several factors and asked how important these factors were in influencing them to hire (n = 3), contract with (n = 27), and/or establish informal arrangements with (n = 11) genetic specialists (Fig. 3). Of the thirty five HMOs reporting at least one of these arrangements, the majority reported the need to offer up-to-date and competitive services, the need to provide cost-effective services, and physician demand as being important factors. Fewer HMOs reported media or public attention, and fear of liability as being important in establishing arrangements with specialists. Six respondents wrote in that genetic specialists were included as part of a larger contract.

Respondents who reported having no arrangement (n = 4) or having informal arrangements with genetic specialists (n = 11) were asked which of the above factors would be important in influencing them to hire or contract with genetic specialists.

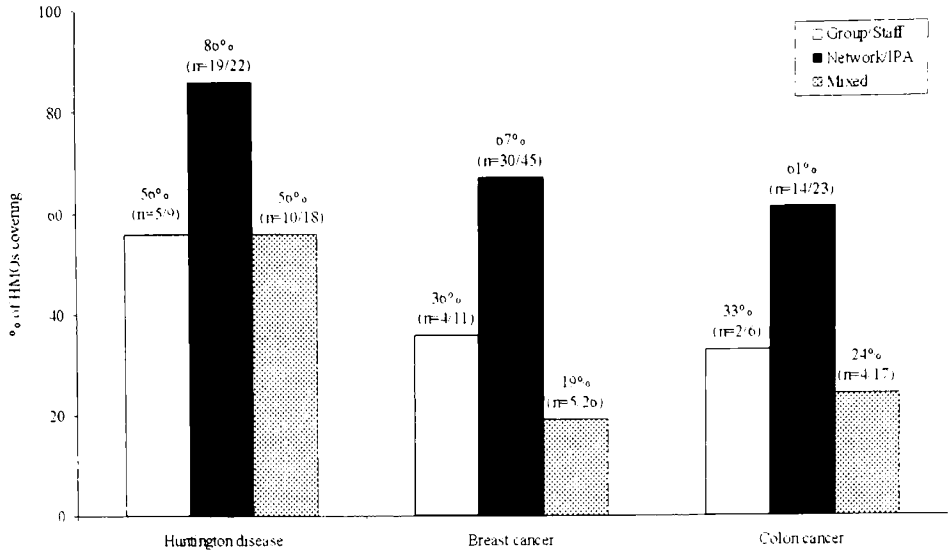

Fig. 1 HMO coverage of requests for predictive testing (written survey).

In their responses, the relative importance of these different factors was the same as for those who had arrangements.

Primary responsibilities of genetic specialists

As part of the phone interview, medical directors were also asked about the primary responsibilities of genetic specialists with whom they contract or employ. The majority (52%, n = 15) reported the primary responsibilities of the genetic specialists as being prenatal and/or pediatric. Twenty five percent (n = 7) reported that they contracted with genetic specialists for "all" types of genetic services. Very few contracted with genetic specialists only for adult medicine or cancer related services.

Education

In the phone interview, we also asked if HMOs had ever developed educational programs or brochures about genetics for nongenetic providers or patients. Seven out of 36 reported that their HMO developed such programs for providers, but only one HMO developed material about genetics for patients. More often the HMO reported that the development of educational materials or programs would be left up to the various clinicians in the HMO.

SURVEY OF DIRECTORS OF ACADEMIC GENETIC CENTER DIRECTORS

Response rate

The response rate was 78% (54/69).

Arrangements with managed care organizations

Seventy-two percent (n = 39) reported that their institution or their genetics unit contracts with managed care organizations. All 15 of the remaining centers reported that they see patients referred by MCOs, although they do not contract with MCOs.

Table 2
Differences in responses to written and phone surveys

	Received requests (n)		Covered test (n)	
	Written	Phone*	Written	Phone*
Breast cancer	35	31	29	21
Colon cancer	24	19	17	11
Alzheimer disease	5	4	1	1

*Confirmed that request/coverage was for predictive genetic testing in unaffected, healthy adults.
n = 39.

Types of services provided

Table 3 describes the number of genetic centers providing initial clinical evaluation, genetic testing, genetic counseling, and ongoing medical management for pediatric, prenatal and adult-onset disorder problems. Eighty percent of genetics units reported providing genetic counseling and 78% provided testing services for adult-onset disorders to patients enrolled in MCOs; whereas only 37% provide ongoing medical management, a significantly smaller proportion than for pediatric (74%) problems (Chi squared, $P < 0.001$).

Corroboration of the relative importance of pediatric and prenatal compared with adult-onset problems was obtained by asking respondents to rank the frequency with which they received referrals from managed care organizations, with 1 being the specialty area for which they received the greatest volume of referrals, 2 the next greatest volume, etc. No respondent ranked adult-onset disorders as generating the highest volume of referrals. The mean rank ± SEM was 1.60 ± 0.11 for prenatal referrals and 1.62 ± 0.10 for pediatric referrals. For adult referrals the mean rank was 2.88 ± 0.09, significantly lower than for prenatal and pediatric referrals (t test, $P < 0.001$). We also gave respondents the opportunity to rank other areas. Three respondents wrote referrals for

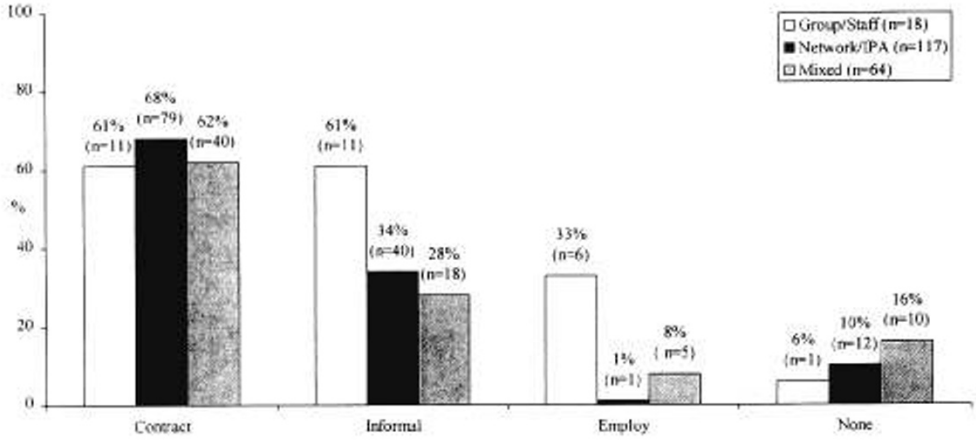

Fig. 2 Types of arrangements with genetic specialists by type of HMO obtained from responses to the written survey. Percentages total to greater than 100, because HMOs could have established more than one arrangement with genetic specialists.

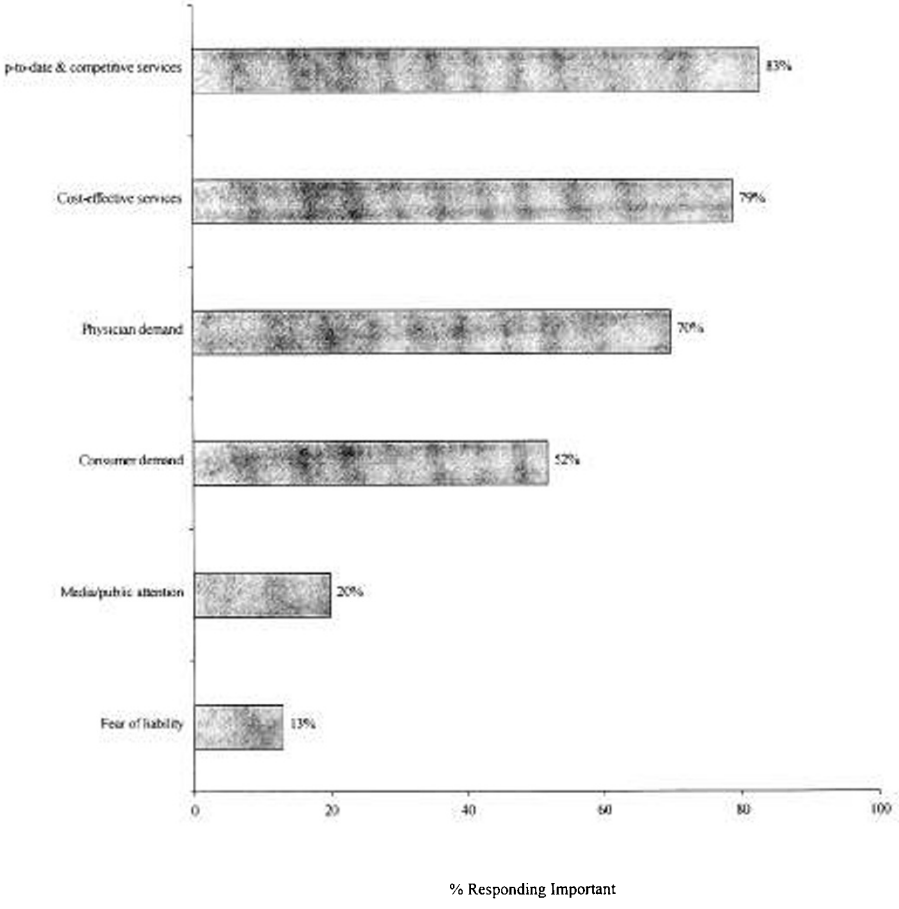

Fig. 3 Importance of factors influencing HMOs to establish arrangements with genetic specialists (n = 35).

laboratory services (mean rank = 2.7) and five each named a different service (mean rank = 3.1).

In order to understand the relative importance of referrals from MCOs for genetic services, directors of genetics units were asked the percentage of patients referred through different sources (Table 4). The highest proportion of patients were referred from independent practitioners, followed by MCOs, and least by patient self-referrals. It is possible that some referrals from "independent" practitioners were from physicians in IPA type HMOs or Point of Service plans. In addition to the above sources of referrals, six directors of genetics units reported referrals through medical assistance programs, ten

Table 3
Services provided by genetic centers to patients covered by managed care organizations

	Pediatric		Prenatal		Adult-onset disorders	
Service	n	%	n	%	n	%
Initial clinical evaluation	50	93	46	85	37	69*
Offering or arranging for genetic tests	51	94	46	85	42	78*
Genetic counseling	51	94	47	87	43	80*
Ongoing medical management	40	74	28	52	20	37*

*One respondent did not answer.

Table 4
Mean percentage of patients referred for genetic services through different sources

Referred by	Mean percent of patients referred (%)
Independent private practitioners	40
Managed care organizations	36
Patient self-referrals	10
Other*:	
Medical assistance (n = 6)	32
State program (n = 10)	30
Academic (n = 3)	25
Other (n = 8)	22

*Write-in responses.
Percentages total to greater than 100 because more than one response was possible.
n = 52.

through state programs, and three through academic institutions or organizations.

Use of laboratories

Directors of genetics units were asked where they send specimens for genetic testing for susceptibility to common complex adult disorders (Table 5). Many centers send specimens to more than one laboratory. Although 39 centers sent some specimens to commercial laboratories, only 4 centers sent all their tests to a commercial laboratory (data not shown).

Table 5
Laboratories used for genetic tests for common, complex adult disorders by genetic centers

Laboratory	n	%*
1. University research laboratory in your hospital	29	55
2. Other hospital/university laboratory	42	79
3. Freestanding commercial laboratory	39	74
4. Other	1	1.8
No laboratory referral	5	9.4

*Percentages add to more than 100 because more than one laboratory is used. For instance, 21 respondents sent specimens to the laboratories listed in the first three rows; 12 to the laboratories listed in rows 2 and 3; and 6 to the laboratories listed in rows 1 and 2.
n = 53.

Coverage issues

Five coverage issues that are anecdotally stated to occur in geneticists' relations with MCOs were listed on the questionnaire. Table 6 shows that nearly all genetic units reported that at least one of these issues had arisen in providing services to patients enrolled in MCOs. Other issues that respondents spontaneously wrote in included genetic evaluation only if the patient is already pregnant, denial of authorization for services, referrals only for the least expensive category of consultation, restrictions due to subcontracts, and problems getting adequate or appropriate referrals. The wording of the question did not permit us to estimate how frequently these issues arose.

Educational programs

Thirty one genetics units (57%) reported that they had developed educational training programs or material for nongenetic health care professionals. The most commonly cited programs were for cancer-susceptibility (n = 18).

Twenty of the units offered CME credits for at least one of their educational programs. Twenty-two units had developed

Table 6
Issues arising in providing services to patients enrolled in managed care organizations

	Yes	
Issue	n	%
Patient authorized for only one visit	49	94
Referral/reimbursement only for patient and not family	50	92
Lack of appropriate referrals	47	89
Lab work not authorized	47	89
No choice of lab	44	83
Other	11	21

Percentages total to greater than 100 because more than one response was possible.
n = 53.

programs or materials for patients. Ten reported having developed cancer-susceptibility programs or materials for patients.

DISCUSSION

HMOs are receiving requests for, and are beginning to cover, predictive genetic testing for common adult-onset conditions. If we assume that one-third of the HMO medical directors who responded to the mail survey mistakenly reported that their company covered breast or colon cancer testing, still 28% of HMOs were covering predictive tests for these conditions in 1996. The majority of those covering predictive testing did so on a case-by-case basis, yet three-quarters of HMOs said they would consider policies regarding predictive testing in the next 5 years. Currently, approximately 94% of insurers (including HMOs) in contact with the major commercial laboratory testing for breast cancer susceptibility cover at least part of the cost of testing, usually on a case-by-case basis, suggesting that genetic susceptibility testing is increasingly become part of medical care.[11]

We did not ask the HMO medical directors which type of specialists saw patients who requested testing for breast or colon cancer. Judging from the responses of academic genetic center directors it seems likely that geneticists are consulted. Eighty percent of genetic center directors said they provided counseling about adult-onset disorders (for which we gave as examples colon cancer and coronary artery disease) and 78% said they were offering or arranging tests for these disorders for patients covered by managed care organizations. This does not mean, however, that most HMO subscribers for whom predictive testing for common disorders is indicated are seen by a geneticist. If they were, it could rapidly become the primary activity of geneticists because of the large numbers of patients at risk for common disorders compared with those at risk for a variety of rare disorders. Providing services for adult-onset disorders was not reported to be the primary activity of geneticists by either the HMOs or the genetic center directors. Both of them reported prenatal and pediatric genetic services as the principal activities of medical geneticists. A much higher proportion of people who may have inherited susceptibility to common, adult-onset disorders may be seen by nongeneticists than by geneticists.

A small survey of managed care organizations in New York suggests that medical directors rely on primary care providers to serve as "gate-keepers" for genetic referrals.[12] Consistent with this was a survey of primary care providers in the Pacific Northwest, in which the majority of the nearly 800 internist and obstetrician/gynecologist respondents said they would provide risk counseling to women at increased risk of breast cancer rather than refer to a specialist.[13] The small and stable supply of geneticists and genetic counselors places limits on the proportion of patients that can be seen by genetic specialists, leading to greater reliance on primary care and other nongeneticist practitioners to provide information on genetic testing.[14,15]

HMOs do have formal relations with medical geneticists. There was agreement in the percentage of HMO medical directors who said they had contractual arrangements with geneticists (65%) and genetic center directors who reported having contractual arrangements with MCOs (72%). The most frequently cited reason HMOs gave for establishing relationships with genetic specialists was providing up-to-date and competitive services. In contrast to our finding, a smaller survey of HMOs in New England reported that only seven of 18 responding HMOs (39%) employed or contracted with medical geneticists or genetic counselors.[16]

Even if most HMOs have formal arrangements with geneticists, as our data suggest, this does not mean that they encourage referral to geneticists. In our survey, the vast majority of genetic center directors encountered some difficulties in MCO coverage, for instance, HMOs would not reimburse for evaluation of extended family members and would only authorize one visit to a geneticist. We do not know how often these problems arose.

Although nongeneticist physicians provide information to patients about genetic susceptibility to adult-onset disorders, they may not be optimally prepared to do so. In the Pacific Northwest study, 14% of internists and obstetrician gynecologists did not know that a young woman with two relatives on her mother's side who had breast cancer was at increased risk and 57% did not know that she would be at increased risk if the same relatives were on her father's side. A majority of those who did not know the risk was increased said they would provide risk counseling.[13] In telephone interviews of physicians who ordered a test for familial adenomatous polyposis, the interpretation of the tests result was incorrect almost one-third of the time (31.6%).[17] In a national survey in which a comprehensive test of knowledge of genetics and genetic tests was mailed to primary care physicians and psychiatrists, 41% of family physicians who did not deliver babies, and 33.1% of internists answered less than two-thirds of the questions correctly. A lower proportion of family physicians who delivered babies (21.4%), obstetrician-gynecologists (23.5%) and pediatricians (13.4%) answered less than two-thirds correctly.[18] That the validity and utility of genetic tests for common disorders often has not been, and legally need not be, established before they become clinically available[1] exacerbates deficiencies in health care providers' knowledge.

Regardless of whether referrals are generated or not, geneticists should educate other health care practitioners about inherited susceptibility and the indications for, as well as the benefits and risks of, predictive genetic tests. Over half of the genetic directors we surveyed, reported that they were developing educational materials on adult disorders for other health practitioners (57%). The most frequently cited category for both groups was cancer susceptibility. It would be useful to examine the content of these materials and the effectiveness of the efforts of genetic centers to improve knowledge of genetics–and related clinical performance–of primary care and other nongeneticist physicians. An analysis of materials made available by clinical laboratories offering genetic tests indicated inaccuracies in information presented to health care providers and patients.[1]

Geneticists were limited by HMOs in the laboratories they could use for genetic testing (Table 6). With biotechnology companies that provide tests staking out patent claims and stringent licensing agreements, it is doubtful that laboratories in academic centers will ever attract referrals for adult-onset disorders as they do for rare, single gene disorders. Almost three-quarters of genetic units used commercial laboratories for some genetic tests for adult-onset disorders. A recent paper expressed alarm that large commercial laboratories were replacing academic laboratories as providers of genetic tests.[19]

These surveys had a number of limitations. We limited the first survey to medical directors of HMOs whereas in our survey of directors of academic genetic centers we asked questions about MCOs in general, although HMOs comprise the largest proportion of managed care organizations. The response rate of 37% in the HMO survey was lower than we expected. We did not include a monetary incentive, primarily because our funds were limited, but we did mail the survey a second time to nonrespondents. In a previous study of medical directors of all types of private health insurers, the inclusion of a $1 bill with a one-page survey yielded a 48% response rate, but enclosure of a check for $10 with a longer follow-up survey only yielded a 37% response rate from HMO medical directors.[2] Variables for which data on nonrespondents were available, did not differ between respondent and nonrespondent HMOs.

We had a better response rate from the directors of the academic genetic units (78%). We cannot, however, generalize to other providers of genetic services, such as private hospitals or free-standing clinics.

A more troublesome concern is the reliability of the written responses to the HMO survey regarding requests for and coverage of predictive genetic tests. In our original script for the telephone follow-up of respondents who said they were covering predictive tests, we did not include a question on requests and coverage for predictive tests. We decided to add this question when we found that a surprisingly large proportion of respondents to the first survey said they were covering predictive tests and that there was a high turn-over rate among medical directors. Efforts to check reliability of mail survey responses are seldom undertaken. Our finding serves as a warning on the reliability of survey responses. We attempted to minimize "wrong" answers in the first, written survey by asking medical directors to forward the questionnaire to someone else in their organization if s/he was better able to answer the questions. It is not clear that this was always done. Eight respondents to the phone survey were not the same person who responded to the written survey. They accounted for only 3 of the 12 responses that changed between the written and phone survey. In considering the implications of our data, we extrapolated from the phone interviews rather than the survey.

CONCLUSIONS

Increasingly, predictive genetic tests for common, complex, adult-onset disorders are being covered by HMOs. Although geneticists are offering these tests, the proportion they provide is unknown and should be the topic of future research. It is doubtful that they provide most of the testing because there are too few genetic specialists available and access to them may be limited by managed care. Nevertheless, geneticists and genetic counselors are more likely to be aware of the tests' limitations than other practitioners whose knowledge of genetics in general and of risks of common disorders in particular is often deficient. In order to ensure appropriate services to people at risk of common disorders either the knowledge of primary care and other nongeneticist practitioners about predictive genetic testing must be improved or access to geneticists increased. Academic genetic centers are providing educational materials to other providers. The ability of these and other approaches to improve practitioners appropriate use of predictive genetic tests needs evaluation.

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The AAHP offers the following definitions for the four model types in its National Directory of HMOs:

Staff: An organized prepaid health care system that delivers health services through a salaried physician group that is employed by the HMO Unit.

Group: An organized prepaid health system that contracts with one independent group practice to provide health services.

Independent Practice Association (IPA): An organized prepaid health care system that contracts directly with physicians in independent practice, with one or more associations of physicians in independent practice, and/or with one or more multi-specialty group practices (but predominantly organized around solo/single specialty practices) to provide health services.

Network: An organized prepaid health system that contracts with two or more independent group practices to provide health services.

APPENDIX

GENETIC SERVICES IN HEALTH MAINTENANCE ORGANIZATIONS (HMOs)

1. Name of organization: _____

2. Approximately what percentage of your HMO's total prepaid enrollment is in each of the following model types?

Network/IPA model _____
Group/Staff model _____
100%

IF YOUR ORGANIZATION HAS DIFFERENT HMO PRODUCTS OR CONTRACTS, ANSWER "YES" TO QUESTIONS 3-5 EVEN IF THE ANSWER APPLIES TO ONLY ONE OF THE PRODUCTS OR CONTRACTS.

3. Please answer parts I, II, and III for **predictive genetic testing of healthy adults** for each of the following disorders.
(Circle the appropriate responses.)

	I. Have you received requests from providers for predictive testing for			II. Have you ever covered this predictive test?			III. For this predictive test, do you have a written policy to			
	Yes	No	Not sure	Yes	No	Not sure	1) cover on provider request?	2) cover after case review?	3) not cover at all?	Not sure
Breast/ovarian cancer	1	2	3	1	2	3	1	2	3	4
Colon cancer	1	2	3	1	2	3	1	2	3	4
Alzheimer disease	1	2	3	1	2	3	1	2	3	4
Huntington disease	1	2	3	1	2	3	1	2	3	4
Other adult disorders (Please list on other side)	1	2	3	1	2	3	1	2	3	4

4. Do you expect your HMO to consider policies, or additional policies, regarding predictive genetic testing for adult-onset disorders

	Yes	No	Not sure
a) in the next year?	1	2	3
b) in the next 5 years?	1	2	3

5. Does your HMO have any of the following arrangements with genetic specialists (M.D. geneticists, genetic counselors, or nurses trained in genetics who provide genetic counseling)? *(Circle all that apply.)*

	Yes	No	Not sure
We employ them	1	2	3
We contract with them	1	2	3
We have informal arrangements with them	1	2	3

6. If there is anyone else in your organization who can give us more information about the provision of predictive genetic testing for adult-onset conditions, please complete the following.

Name _____ Position _____
Phone _____ email _____

7. What is your name? _____ Position _____

8. Would you like to receive the results of this study?

Yes	No
1	2

Thank you for taking the time to complete this survey. Please return it in the enclosed self-addressed stamped envelope to Genetics & Public Policy Studies, 550 North Broadway, Suite 511, Baltimore, MD 21205-2004.

PROVISION OF GENETIC SERVICES FOR NON-GENETICS SPECIALTIES AND MANAGED CARE ORGANIZATIONS

Respondent's name: _____

Respondent's title: _____

Name of Institution: _____

Name of Genetics Unit or Department: _____

1. Does the genetics unit at your institution **contract** with any managed care organizations? (Managed care organizations include, but are not limited to, health maintenance organizations, independent practice associations, preferred provider organizations, and employer plans). *(Circle the correct number.)*

Yes	No
1	2

2. If the unit does **not** contract with any managed care organizations, does the genetics unit see patients **referred** by managed care organizations or by physicians within managed care organizations?

Yes	No
1	2

If you answered yes to either question 1 or 2, please answer questions 3 and 4. If you answered "no" to both 1 and 2, please go to question 5.

3. What types of genetic services does the genetics unit provide to patients covered by managed care organizations? *(Circle all numbers that apply.)*

Service	Pediatric	Prenatal	Adult-onset disorders (e.g. colon cancer, coronary artery disease, etc).
Initial clinical evaluation	1	2	3
Offering or arranging for genetic tests	1	2	3
Genetic counseling	1	2	3
Ongoing medical management	1	2	3

4. Please rank the frequency with which the genetics unit receives referrals from managed care organizations in each of the areas listed below. The area for which the greatest volume of patients are referred should receive a ranking of 1, the area for which the least volume of patients are referred should receive a ranking of 4, 5, or 6, depending on the services the unit provides.

Prenatal ..... _____

Pediatrics ..... _____

Adult-onset ..... _____

Other (Please specify _____) ..... _____

Other (Please specify _____) ..... _____

Other (Please specify _____) ..... _____

5. Approximately what percentage of patients seen in the genetics unit are referred for genetic services through each of the following:

a. independent private practitioners (not through a managed care organization) ___%

b. patient self-referrals ___%

c. managed care organizations or providers in managed care organization ___%

d. Other_____ ___%

Total 1 0 0%

6. Do any genetic specialists (M.D. geneticists, genetic counselors, or nurses trained in genetics) in the unit provide genetic services at **non-genetic** specialty sites (**not** limited to managed care organizations)?

	Yes	No
	1	2

If yes, what sites? *(Circle all that apply.)*

a. Birth defects 1
b. Cardiology 2
c. Endocrinology 3
d. Hematology 4
e. Neurology/Psychiatry 5
f. Obstetrics/Gynecology 6
g. Oncology 7
h. Pulmonary 8
i. other (specify) 9

7. Where does the genetics unit send specimens for genetic testing for susceptibility to common complex adult disorders? *(Circle all that apply.)*

a. University research laboratory in your hospital 1

b. Other hospital/university laboratory 2

c. Freestanding commercial laboratory 3

d. Have not performed genetic susceptibility testing 4

e. Other (Please specify) _____ 5

8. Which of the following are involved in deciding to offer a new genetic testing service (including offering or arranging for genetic testing, education, and counseling)? *(Circle all that apply.)*

a. Director of genetics unit 1

b. M.D. geneticist or genetic counselor himself/herself 2

c. Director of cytogenetics or molecular biology laboratory 3

d. Other specialty clinic 4

e. Executive director of hospital/institution 5

f. Advisory committee (Please describe) _____ 6

g. Provided at patient request 7

h. Other (Please specify) _____ 8

9. Has the genetics unit had any of the following or other problems providing services to patients enrolled in managed care organizations?

	Yes	No
Patient authorized for only one visit	1	2
Referral/reimbursement only for patient and not extended family	1	2
Lack of appropriate referrals	1	2
Laboratory work not authorized	1	2
No choice of laboratory	1	2

Other: _____

10. Has the genetics unit developed any educational training programs (workshops, conferences, etc.) or any educational materials (pamphlets, fact sheets, videos, etc) about adult-onset disorders...?

A. For non-genetic health care professionals (family practitioners, internists, obstetricians/gynecologists, social workers, midwives, nurses)?

Yes	No	Not sure
1	2	3

1. If yes, please list the subject(s) of the program(s).

_____

_____

_____

2. Were continuing education credits available?

Yes	No	Not sure
1	2	3

B. For patients?

Yes	No	Not sure
1	2	3

1. If yes, please list the subject(s) of the program(s).

_____

_____

_____

11. Would you like to receive a summary of the results of our studies?

Yes	No
1	2

Thank you for taking the time to complete this survey. Please return it in the enclosed self-addressed stamped envelope to:

Genetics & Public Policy Studies
The Johns Hopkins Medical Institutions
550 N. Broadway, Suite 511
Baltimore, MD 21205-2004