Coverage and Reimbursement of Genetic Tests and Services

Report of the
Secretary's Advisory Committee on Genetics, Health, and Society

February 2006

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DEPARTMENT OF HEALTH & HUMAN SERVICES Public Health Service

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February 1, 2006

The Honorable Michael O. Leavitt
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Leavitt:

In keeping with the mandate of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) to serve as a public forum for deliberations on the broad range of human health and societal issues raised by the development and use of genetic technologies and, as warranted, to provide advice on these issues, I am pleased to submit this report on *Coverage and Reimbursement of Genetic Tests and Services*. The report, which is the culmination of more than a year of fact-finding, consultation and deliberation by the Committee, describes the current state of coverage and reimbursement of genetic tests and services, highlights how problems in the system are affecting patient access to tests and services, and identifies nine steps that can be taken to overcome the barriers to patient access.

Assessing how genetic tests are being integrated into health care and public health has been a primary focus of the Committee's work since its inception in 2003. In its first year, SACGHS identified coverage and reimbursement as a high priority issue warranting in-depth deliberation and analysis because of its importance in ensuring appropriate access to and clinical integration of genetic tests and services. Through consultations with various experts and members of the public, the Committee identified significant barriers and unmet data needs that are limiting appropriate access to and clinical integration of genetic tests and services.

The nine recommendations contained in this report identify steps that the Department of Health and Human Services could take to help improve appropriate access to and utilization of health-related genetic tests and services in both public and private health insurance programs. We believe that these changes are critical to the integration of genetic tests and services into the health care system and in the long run will help the Department fulfill its mission to improve the health and well-being of Americans.

Sincerely,

Reed V. Tuckson, M.D.
Chair, SACGHS

About SACGHS

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) was chartered in 2002 by the Secretary of Health and Human Services (HHS) as a public forum for deliberations on the broad range of human health and societal issues raised by the development and use of genetic tests and, as warranted, to provide advice on these issues. The charter sets outs the following specific functions of the Committee:

- Assessing how genetic tests are being integrated into health care and public health;
- Studying the clinical, ethical, legal, and societal implications of new medical applications, such as preimplantation genetic diagnosis, and emerging technological approaches to clinical testing;
- Identifying opportunities and gaps in research and data collection efforts;
- Exploring the use of genetics in bioterrorism;
- Examining current patent policy and licensing practices for their impact on access to genetic tests;
- Analyzing the uses of genetic information in education, employment, insurance (including health, disability, long-term care, and life), and law (including family, immigration, and forensics); and
- Serving as a public forum for the discussion of emerging scientific, ethical, legal, and social issues raised by genetic tests.

Structurally, SACGHS consists of 13 individuals from around the Nation who have expertise in disciplines relevant to genetics and genetic technologies. These disciplines include molecular biology, human genetics, health care, public health, bioterrorism, ethics, forensics, law, psychology, social sciences, education, occupational health, insurance, and consumer issues, among others. At least 2 of the 13 members must have expertise in consumer issues or an understanding of the views and perspectives of the general public.

Representatives of 19 Federal department or agencies also sit on SACGHS in an ex officio (nonvoting) capacity. The departments and agencies are the Department of Commerce, Department of Defense, Department of Education, Department of Energy, Administration for Children and Families (HHS), Agency for Healthcare Research and Quality (HHS), Centers for Disease Control and Prevention (HHS), Centers for Medicare & Medicaid Services (HHS), Food and Drug Administration (HHS), Health Resources and Services Administration (HHS), National Institutes of Health (HHS), Office for Civil Rights (HHS), Office for Human Research Protections (HHS), Office of the Secretary (HHS), Department of Justice, Department of Labor, Department of Veterans Affairs, Equal Employment Opportunity Commission, and Federal Trade Commission.

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Acknowledgments

The Committee wishes to thank the members of the SACGHS Coverage and Reimbursement Task Force for their pivotal role in guiding the development of this report. The Task Force was chaired by Cynthia Berry and composed of Debra Leonard, Reed Tuckson, Emily Winn-Deen, Francis Chesley, Muin Khoury, Steve Phurrough, James Rollins, and Marc Williams.

The Committee is indebted to the following for sharing their knowledge and expertise on the issues surrounding the coverage and reimbursement of genetic tests and services:

- Ronald Bachman (Kaiser Permanente Northern California), Andrea Ferreira-Gonzalez (Virginia Commonwealth University Medical Center), Michele Schoonmaker (Cepheid), Don Thompson (CMS), Sean Tunis (CMS), David Veenstra (University of California-San Francisco), and Marc Williams (IHC Clinical Genetics Institute) for their presentations and participation in a roundtable discussion with SACGHS at the March 2004 meeting.
- Paul Billings (LabCorp), Linda Bradley (CDC), Tammy Karnes (LabCorp), Terrence Kay (CMS), Joe Kelly (CMS), and Don Thompson (CMS) for their participation in the September 2004 Task Force meeting.

The Committee thanks all of the individuals and organizations who responded to the Committee's requests for public comments during the development of this report (see Appendix C). The Committee gave careful consideration to each of the comments. The input has enhanced the report's analysis and the cogency of the recommendations.

The Committee wishes to recognize the work of SACGHS staff. Suzanne Goodwin and Amanda Sarata were lead staff members on the project, responsible for organizing the deliberations of the Task Force and full Committee and drafting the report. In addition, Fay Shamanski worked on the discussion of genetic/genomic tests and technologies. Sarah Carr provided guidance to the staff. The Committee thanks the NIH Office of Biotechnology Activities, under the direction of Amy Patterson, for its ongoing support and operational management of SACGHS.

Contents

Preface

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) was chartered in 2002 to provide advice to the Secretary of Health and Human Services on human health and societal issues raised by the development and use of genetic tests. Because the scope of its charge encompasses a broad range of issues, the Committee undertook a prioritization process during its first year to help focus its attention on areas in which policy recommendations will have the greatest impact on the integration of genetics and genomics into health care. The Committee ranked coverage and reimbursement of genetic tests and services as a high-priority issue warranting indepth deliberation and analysis. The Committee concluded that coverage and reimbursement issues are critical to ensuring appropriate access to genetic tests and services and their integration into clinical practice. SACGHS also realized that significant barriers and unmet data needs are limiting appropriate access and clinical integration and that the Department of Health and Human Services can take steps to minimize or eliminate these barriers.[1] This report is the culmination of SACGHS's yearlong study of the issue.

SACGHS believes that coverage and reimbursement are critical to ensuring appropriate access to genetic tests and services and their integration into clinical practice, that there currently exist significant barriers and unmet data needs that are limiting appropriate access and clinical integration, and that the Department of Health and Human Services can be influential in minimizing or eliminating these barriers.

The Committee began its deliberations on coverage and reimbursement issues at its third meeting in March 2004 by convening a panel of experts. The panel was made up of two clinical geneticists, an academic laboratorian, a pharmacoeconomist, an expert on the coverage of genetic tests, and two officials from the Centers for Medicare & Medicaid Services. The background and data presented by these experts gave the Committee an overview of the relevant aspects of coverage and reimbursement that relate to genetic tests and services as well as barriers and gaps that are impeding appropriate access.

SACGHS also assembled a Task Force comprised of Committee members and ex officio members to guide the staff in the development of the report. The Task Force, chaired by Cynthia Berry, held a 1-day meeting in September 2004 between Committee meetings and convened a number of times by conference call and e-mail.

In addition, the public had opportunities to inform and provide input on the development of this report. At each meeting, copies of the draft reports were made available, time was provided for oral testimony, and written comments were encouraged. Also, a Federal Register notice was published in April 2005 soliciting public comment on the penultimate draft of the report. In response to this request, the Committee received comments from 83 organizations and individuals (see Appendix C).

[1] A Roadmap for the Integration of Genetics and Genomics into Health and Society: The Study Priorities of the Secretary's Advisory Committee on Genetics, Health, and Society. June 2004. http://www4.od.nih.gov/oba/sacghs/reports/SACGHSPriorities.pdf [November 18, 2005].

Executive Summary

Although advances in genetics and genomics are driving the development of new genetic tests and services, problems with coverage and reimbursement are limiting their accessibility and integration into the health care system. This report describes the state of coverage and reimbursement of genetic tests and services and problems and barriers in the system. To address the problems, the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) makes the following recommendations to alleviate the barriers and improve current mechanisms for coverage and reimbursement of genetic tests and services.

Recommendation 1: Evidence-Based Coverage Decisionmaking

Health care cost constraints, demands to improve health outcomes, a greater emphasis on quality, and the introduction of new technologies and procedures available for clinical use all are driving health care payers to reassess how they make decisions about which tests and services to cover and under what conditions they will reimburse them. Health insurance plans have emphasized evidence-based coverage decision making as a way to determine which technologies and services are appropriate to cover. However, the evidence needed to make informed coverage decisions is lacking for many genetic tests and services. In addition, many genetic diseases are rare and/or currently lack therapeutic and preventive options, and rationalizing coverage for genetic tests and services can be more difficult when short-run costs cannot be recouped in short timeframe.

1. **The Secretary should task an appropriate group to develop a set of principles to guide coverage decision-making for genetic tests and services. The guiding principles should address the issues identified in this report, including economic evaluation/cost-effectiveness, prevention, rare disease tests, therapeutic benefit, and informational utility. The group also should assess the type, quality, and quantity of existing evidence for specific genetic tests to determine whether the evidence is adequate to establish a test's analytical validity, clinical validity, and clinical utility. If not, the group should identify any evidentiary gaps.**

   This group should consist of experts from both the public (i.e., U.S. Department of Health and Human Services [HHS] agencies) and private sectors and make use of resources and models from both sectors. A work group organized by the Centers for Disease Control and Prevention, called the Evaluation of Genomic Applications in Practice and Prevention Working Group, is an example of such a group. It is made up of a diverse range of experts from both sectors and is performing related work, and thus could be tasked to develop these principles.

   In addition, a mechanism should be established to promote and fund studies to address evidentiary gaps identified by the group.

Recommendation 2:
Medicare's Influence on the Private Insurance Market

Because Medicare is the largest provider of health insurance in the United States, its coverage decisions are closely monitored by private health insurance plans. Because genetic tests often are used for preventive, reproductive, or life planning purposes and because most hereditary diseases will manifest prior to age 65, it may not be appropriate for private health insurance plans to follow Medicare's lead in making coverage decisions for predictive and predispositional genetic tests and services.

2. **Although standardization of coverage decisions using the best scientific evidence across public and private payers is ideal (see Recommendation 1), private health insurance plans are encouraged to make their own coverage determinations about genetic tests and services relative to the populations they serve. The group described in Recommendation 1 should make available the scientific evidence needed to make these decisions.**

Recommendation 3:
Medicare Coverage Decisionmaking Process

Medicare coverage decisions are made at both the national and local levels. Although national decisions apply to all beneficiaries, local decisions apply only to those beneficiaries living in the particular region (28 regions in all), which can lead to inconsistencies in coverage from one region to another. This dual system impedes rapid and widespread coverage of genetic tests and services.

3. **The Secretary should encourage the Centers for Medicare & Medicaid Services (CMS) to move forward with the development of a plan to evaluate new local coverage decisions to determine which should be adopted nationally and to what extent greater consistency in Medicare coverage policy can be achieved (such a plan is mandated in Section 731 of the Medicare Prescription Drug, Improvement, and Modernization Act of 2003). As part of its implementation of Section 731, CMS should consider a mechanism that would automatically initiate a national coverage review process for any test or service approved for coverage by a certain number of local Medicare administrative contractors.**

Recommendation 4:
Medicare Screening Exclusion

Federal statute prevents Medicare from covering preventive services unless explicitly authorized by Congress. The screening exclusion is embodied in a CMS policy that states, "Tests for screening purposes that are performed in the absence of signs, symptoms, complaints, or personal history of disease or injury are not covered except as explicitly authorized by statute." Since CMS considers predictive and predispositional genetic tests to be screening tests and since coverage of such tests has not been explicitly authorized by Congress, they are not covered by Medicare. The screening exclusion also limits Medicare coverage for genetic counseling.

4. **Predictive and predispositional genetic tests can be clinically beneficial even when there are no current signs, symptoms, or personal history of illness. As such, predictive and predispositional genetic tests and their accompanying services that meet evidence standards should be covered under Medicare.**

The Secretary should urge Congress to add a benefit category for preventive services that would enable CMS to use its national coverage decisionmaking process, which includes an assessment of existing evidence, to determine whether a test or service is reasonable and necessary for the prevention or early detection of an illness or disability in asymptomatic individuals and, thus, ought to be covered. A statutory change would allow CMS to consider covering many more genetic tests and services used for preventive purposes.

More immediately, the Secretary should direct CMS to clarify that, in certain cases as scientific evidence warrants, a "personal history" of disease can include having a family history of a disease. This change would make it possible for a beneficiary with a family history of a disease to meet the "reasonable and necessary" standard for Medicare coverage. CMS will need to develop criteria that define when a family history should be considered a personal history of disease.

Recommendation 5: Medicaid Coverage of Genetic Tests and Services

With the exception of newborn screening, genetic tests and services are optional Medicaid benefits. As a result, coverage for genetic tests and services can be affected by State budget cuts. Changes in States' Medicaid funding can create instability in access to genetic tests and services for the Medicaid population. Also, variation in Medicaid coverage across States can result in disparate access to genetic tests and services. Information and resources for making informed Medicaid coverage decisions can help minimize State variation in access to genetic tests and services.

5. **The Secretary should ensure that States receive information about the existing evidence base and other supporting information about genetic tests and services, such as guiding principles that serve as the basis for coverage decisionmaking (see Recommendation 1). This information should be used by States to inform their Medicaid coverage decisions.**

   Through the provision of grant funding, HHS should continue to encourage States to cover, adopt, and provide genetic tests and services with a sound evidence base.

Recommendation 6: Medicare Clinical Laboratory Fee Schedule

Many providers who bill Medicare contend that Medicare's payment rates for clinical laboratory tests have not kept pace with inflation or with economic and technological changes in laboratory practices. As a result, Medicare laboratory fees often do not reflect a genetic test's true cost. In addition, Congress imposed a freeze on payment rates for clinical laboratory tests, locking rates at the 2003 level until 2009. HHS does have authority (known as the "inherently reasonableness" authority) to revise payment levels when they threaten beneficiary access to care or represent a misappropriation of taxpayer dollars.

6. **When the congressional freeze on laboratory payment rates ends in 2009, the Secretary should be prepared to revise payment rates to reflect the true cost of a genetic test. In the meantime, the Secretary should direct CMS to invoke its inherent reasonableness authority to address variations in payment rates for the genetic test Current Procedural Terminology (CPT) codes.**

Recommendation 7:
Billing and Reimbursement for Genetic Counseling Services

Although genetic counseling is often critical to ensuring the appropriate use of genetic tests, counseling services are not being adequately reimbursed, a situation that can lead to access problems for patients in need of such services. The reimbursement problem has at least three sources. First, current CPT codes for billing are inadequate. Genetic counseling sessions can last for 2 to 3 hours, but the highest available CPT code accounts for a significantly shorter timeframe. There is a way to augment the CPT code, but such prolonged service codes are rarely reimbursed. Second, not all genetic counseling providers are eligible to bill Medicare directly. Currently, nurse practitioners, physician assistants, certified nurse specialists, certified nurse midwives, clinical psychologists, and clinical social workers are statutorily eligible to bill Medicare directly. Other nonphysician counseling providers must bill "incident to" a physician and when billing Medicare for their services, they may use only certain CPT codes. Third, State licensure is an important credential for being recognized by payers as a qualified provider, yet only three States have authorized the licensing of genetic counselors.

7a. To ensure full access to genetic counseling services for all Americans, the Secretary should expeditiously identify an appropriate entity to determine (1) which health professions are qualified to provide genetic counseling services (see page 49 for discussion of genetic counseling services and providers), and of those determined to be qualified, (2) which should be able to practice without physician supervision and, thereby, bill payers directly for their services. The entity selected to make these determinations should be guided by the professions' credentials, licensure status, scope of practice, and any other criteria deemed appropriate. The credentialing standards of a number of professional societies, such as the American Board of Genetic Counseling and the Genetic Nursing Credentialing Commission, could be used as a reference point. A description of existing credentialing programs is provided in Appendix B.

If this review process results in the determination that a health profession should be allowed to practice independently, the Secretary should urge Congress to add this health profession to the list of nonphysician practitioners eligible to bill Medicare directly for their services.

7b. HHS should assess the adequacy of existing CPT Evaluation & Management (E&M) codes and their associated relative values with respect to genetic counseling services. This assessment should be carried out with input from genetic counseling service providers. HHS should address any inadequacies as deemed appropriate.

7c. The Secretary should direct CMS to allow nonphysician health providers who are deemed qualified to provide genetic counseling services and who currently bill incident to a physician to use the full range of CPT E&M codes available for genetic counseling services.

7d. The Secretary should ensure that all HHS programs are reimbursing prolonged service codes when they are determined to be reasonable and necessary.

7e. The Secretary should direct CMS to deem all nonphysician health providers permitted to bill a health plan directly as eligible for a National Provider Identifier.

Recommendation 8:
Provider Education and Training

Genetic tests are being marketed to health providers and directly to consumers. If providers are not adequately trained in the use and interpretation of genetic tests, they may provide inappropriate services to their patients and expect to be reimbursed for them. Providers need adequate genetics education and training to know when genetic tests are appropriate and to help their patients make decisions about when to be tested. A working knowledge of genetics also is important for health payers because it will help them make informed and appropriate coverage decisions.

8. **Since genetic tests and services are being integrated into all areas of health care and since providers have an important role in ensuring appropriate use of and access to genetic tests and services among diverse populations, there is a critical need for programs to educate and train health providers and payers in genetics and genomics. Health providers should be able to meet established genetic competencies and, thereby, integrate genetics effectively into their practices. The Secretary should develop a plan for HHS agencies to work collaboratively with Federal, State, and private organizations to develop, catalog, and disseminate case studies and practice models that demonstrate the relevance of genetics and genomics.[2]**

   The Secretary should provide financial support to assess the impact of genetics education and training on health outcomes.

   The Secretary should strive to incorporate genetics and genomics into relevant initiatives of HHS, including the National Health Information Infrastructure.

Recommendation 9:
Public Awareness

Public awareness of new health care tests and treatments can create consumer demand. Although greater public awareness and demand can facilitate coverage for new, safe, efficacious, and appropriate genetic tests and services, because of the complexity of genetic tests, they also can result in misinformation and inappropriate demand for genetic tests and services.

9. **For patients and consumers to evaluate health plan benefits and health providers and make the most appropriate decisions for themselves and their families, they need reliable and trustworthy information about family history, genetics, and genetic technologies. The Secretary should ensure that educational resources are widely available through Federal Government Web sites and other appropriate public information mechanisms to inform decisions about genetic tests and services.**

Implementation of these recommendations should help improve appropriate access to and utilization of genetic tests and services by ensuring appropriate coverage and reimbursement throughout the health care system. Although the recommendations are primarily directed at HHS, the Committee hopes that private health insurance plans also will address the identified barriers that are relevant to them.

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[2] SACGHS Resolution on Genetics Education and Training of Health Professionals, June 2004. http://www4.od.nih.gov/oba/sacghs/reports/EducationResolutionJune04.pdf [December 14, 2005].

Introduction

Scientific and technical advances have expanded our knowledge of the genetic contributions to disease and have made possible the development of genetic tests that are capable of diagnosing current disease, assessing the risk of future disease, and enabling treatment to be tailored to individual genetic variations. The recent completion of the sequencing of the human genome is increasing the pace of these advancements, all of which promise further improvements in the diagnosis, treatment, and prevention of disease, more informed decisionmaking, better disease management, and more efficient use of health care resources.

Although advances in genetics and genomics are driving the development of new genetic tests and services, problems with coverage and reimbursement of current genetic tests and services are limiting their accessibility and integration into the health care system.[3] These problems include inadequate data to support evidence-based coverage decisions, the lack of a uniform and broadly accepted process for identifying and addressing gaps in evidence, and limitations in the Medicare program.

While addressing these problems, it is important to consider the larger context in which these tests and services are provided. The U.S. health care system currently faces numerous cost, quality, and access challenges, each of which affects the appropriate integration of genetic tests and services into clinical and public health practice. For example, 46 million Americans are uninsured;[4] between 44,000 and 98,000 fatal, preventable medical errors occur each year;[5] only 55 percent of health care follows with evidence-based guidelines;[6] and health care costs are escalating by approximately $120 billion a year.[7] In addition, the health care system is overwhelmed by the task of integrating new technologies, pharmaceuticals, and procedures generated by biomedical research.[8] In light of these broader system challenges, problems associated with the introduction, delivery, and financing of genetic tests and services should be kept in perspective and considered with great care. Although appropriate access to genetic tests and services has the potential to facilitate improvements in health status, efforts to address limitations in coverage and reimbursement of genetic tests and services should be balanced against the constraints of limited and finite resources and responsible allocation of health care dollars.

This report has two main purposes: (1) to describe the current state of, and problems associated with, coverage and reimbursement of genetic tests and services and (2) to offer recommendations on how current

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[3] Public comments to SACGHS on draft report on Coverage and Reimbursement of Genetic Tests and Services. June 2005.
[4] U.S. Census Bureau. Income, Poverty, and Health Insurance Coverage in the United States: 2003. http://www.census.gov/prod/2004pubs/p60-226.pdf [November 18, 2005].
[5] Kohn LT, Corrigan JM, and Donaldson MS, eds. To Err Is Human: Building a Safer Health System. Washington DC: National Academies Press, 2000. http://www.nap.edu/books/0309068371/html/ [November 18, 2005].
[6] McGlynn EA et al. The quality of health care delivered to adults in the United States. *NEJM* 2003. 348(26):2635-45.
[7] Centers for Medicare & Medicaid Services, Office of the Actuary, National Health Statistics Group. National Health Expenditures Aggregate Amounts and Average Annual Percent Change, by Type of Expenditure: Selected Calendar Years 1980-2003. http://new.cms.hhs.gov/NationalHealthExpendData/downloads/nhetables.pdf [January 4, 2006].
[8] Institute of Medicine Committee on the Quality of Health Care in America. Crossing the Quality Chasm: A New Health System for the 21st Century. Washington, DC: National Academies Press, 2001. http://www.nap.edu/books/0309072808/html/ [November 18, 2005].

This report makes nine recommendations to improve appropriate access to and utilization of health-related genetic tests and services by ensuring appropriate coverage and reimbursement throughout the health care system.

It should be noted, however, that SACGHS does not advocate coverage of all genetic tests and services under all circumstances. Rather, the Committee believes that genetic tests and services should be covered when there is adequate evidence to support their use. In addition, the Committee believes that reimbursement levels for covered tests should be set at levels that do not undermine coverage or reduce appropriate patient access.

mechanisms for coverage and reimbursement of genetic tests and services might be improved. The report makes nine recommendations to improve appropriate access to and utilization of health-related genetic tests and services by ensuring appropriate coverage and reimbursement throughout the health care system. It should be noted, however, that the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) does not advocate coverage of all genetic tests and services under all circumstances. Rather, the Committee believes that genetic tests and services should be covered when there is adequate evidence to support their use. In addition, the Committee believes that reimbursement levels for covered tests should be set at levels that do not undermine coverage or reduce appropriate patient access. Other considerations, such as the test's benefits, risks, and cost; the subsequent costs involved in responding to the test results; and patient preferences, also need to be factored into the decisionmaking process.

This report does not intend to address general problems with access to health care in the United States (e.g., uninsured and underinsured individuals or disparities in health and health care). Other groups with broader mandates and expertise are studying and seeking solutions to these more global problems. Also, although intending to address only problems specific to coverage and reimbursement of genetic tests and services, the Committee's recommendations may have broader implications and, if implemented, could affect other areas of health care. The Committee also realizes that implementing the nine recommendations outlined in this report will not eliminate other barriers to access, such as the public's fear of genetic discrimination (see page 11).

Although the Committee's mandate pertains to genetic technologies broadly, for the purposes of this report, SACGHS is focusing on genetic tests and services that are currently in clinical use. Many of the issues discussed in this report may, nonetheless, be applicable to genetic and genomic technologies now in development.

Finally, although the Committee's recommendations are primarily directed to HHS, many of the identified barriers (e.g., underpayment of genetic tests and services) also are applicable to private health insurance plans. The Committee hopes that private plans will address relevant barriers in a suitable manner.

Genetic Discrimination

Genetic discrimination is the potential use of genetic information to discriminate against people in the workplace and in health insurance. Even if the financial barriers to access identified in this report were eliminated, beneficial genetic tests and services may continue to be underutilized due to the public's fear of genetic discrimination.

Health insurance organizations and groups representing employers do not believe that insurers or employers are currently engaging in genetic discrimination. Although genetic discrimination does not appear to be a widespread problem at this time, there have been a number of cases that have been reported prominently in the media. Also, surveys indicate that people are fearful that health insurers and employers will misuse presymptomatic genetic information, and some forego genetic testing because of this fear.[1-4]

Current Federal and State laws in this area are considered inadequate,[5] and Federal legislation prohibiting the misuse of genetic information is needed to address remaining gaps. In June 2003, March 2004, and May 2005, SACGHS wrote to the Secretary of Health and Human Services to express support for Federal genetic nondiscrimination legislation. The Genetic Information Nondiscrimination Act of 2005 (S.306) was approved by the U.S. Senate in February 2005. The Administration issued a Statement of Administration Policy expressing support for the passage of S.306. In March 2005 a bipartisan companion bill (H.R. 1227) was introduced in the U.S. House of Representatives.

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[1] Secretary's Advisory Committee on Genetic Testing. Public Consultation on Genetic Testing, January 27, 2000. http://www4.od.nih.gov/oba/sacgt/transcripts/sacgttran1-00.pdf [November 18, 2005].
[2] Secretary's Advisory Committee on Genetics, Health, and Society. Genetic Discrimination Session, October 18, 2004.
[3] Coalition for Genetic Fairness. Faces of Genetic Discrimination. 2004. http://www.nationalpartnership.org/portals/p3/library/geneticdiscrimination/facesofgeneticdiscrimination.pdf [November 18, 2005].
[4] Hadley DW et al. Genetic counseling and testing in families with hereditary nonpolyposis colorectal cancer. *Arch Intern Med* 2003. 163:573-582.
[5] Lanman RB. An Analysis of the Adequacy of Current Law Protecting against Genetic Discrimination in Health Insurance and Employment. May 2005. http://www4.od.nih.gov/oba/sacghs/reports/legal_analysis_May2005.pdf [November 18, 2005].

Genetic Tests & Services: Challenges to the U.S. Health Care System

Genetic tests are like other laboratory services in that they detect biological products and analytes and can provide diagnostic information that informs clinical treatment decisions. In other ways, these new technologies and services have expanded and challenged our previous understanding of the role of laboratory services in health care. Genetic tests and services are different (but not necessarily unique) in that they are relevant to all clinical disciplines and because they can provide more information than traditional laboratory tests. In addition to providing diagnostic information, genetic tests have the potential to provide more precise, accurate information about an individual's susceptibility to disease and response to pharmaceuticals. Furthermore, because genes are inherited, genetic tests can clarify family history and have implications for other family members. Since an individual's heredity does not change over time, a specific genetic test has to be performed only once in a lifetime.[9] For these reasons, genetic tests and services have broadened the diagnostic and predictive capabilities of clinical laboratories and, in some cases, replaced older methods of diagnosis.

Genetic tests and services face many of the same challenges that other new medical technologies encounter as they become integrated into the health care system, such as building a sufficient evidence base that demonstrates that they are similar or superior to existing technologies and services. Some of the same processes that have successfully allowed for the integration of other new medical technologies can help with the integration of genetic tests and services. Although genetic tests share many of the same difficulties as other laboratory tests, they also have some features that pose additional challenges to the current health care financing and delivery system.

First, genetic tests may require the involvement of a team of providers—a primary care provider to coordinate the patient's health care, a medical geneticist to assist with the diagnosis and treatment of disease, a genetic counseling provider to educate and counsel the patient before and after genetic testing, a laboratorian to carry out testing, a specialist to manage treatment, and one or more allied health professionals to provide any additional social support services. Inadequate coverage and reimbursement of each provider's services can affect their ability to provide these services and the overall quality of the patient's care is diminished.

[9] A genetic test that identifies other mutations not previously tested for is considered to be different from the original test. Also, if new information becomes available (e.g., a mutation is found to be associated with another condition), the original test should not need to be repeated since the original test results can be reinterpreted in light of the new information. However, genetic tests that measure gene expression or identify somatic mutations may need to be repeated more than once in a person's lifetime.

Second, many diseases are often the result of the interaction of more than one risk factor. Genetic mutations or variations are one type of risk factor for complex diseases. No clinical interventions are currently available to modify the genetic mutation (although gene transfer strategies may make this possible in the future). However, genetic testing can identify genetic risks and provide individuals with knowledge that can be used to reduce their risks (e.g., lifestyle changes, increased surveillance, prophylactic interventions) that might not otherwise have been taken had genetic testing not been performed. For instance, women who test positive for BRCA1 or BRCA2 mutations for breast cancer may choose to obtain mammograms more frequently or at an earlier age than recommended for the general population, or they may decide to have a prophylactic mastectomy and/or prophylactic oophorectomy to reduce their chances of developing breast and ovarian cancer. These steps do not eliminate the genetic mutation, but they reduce the overall risk of disease.

Third, some of the diseases for which genetic tests are available for risk assessment have no treatments or clinical interventions. For example, individuals who test positive for Huntington disease have close to a 100 percent risk of developing the disease. However, there is no available therapy that can prevent onset or alleviate symptoms. Although there are no therapeutic options available, this information may still be useful for differential diagnosis and overall clinical management. Furthermore, for many people, knowledge of their risks can be useful for family and long-term care planning purposes as well as psychosocial well-being.

Fourth, unlike many other laboratory tests, genetic tests raise complex legal, ethical, societal, psychological, familial, and personal issues. For example, genetic testing can raise concerns about privacy and confidentiality, genetic discrimination, reproductive options, social stigmatization, and personal and group identity. In addition, genetic research may result in new knowledge that has implications for an individual based on previous test results. Because of these complexities, genetic counseling often is warranted to ensure that individuals are informed of the implications of their testing decisions and the limitations of the results.

Finally, in the case of genetic tests for heritable mutations, an individual's test results can have implications for other blood relatives. In some cases, other family members may need to be tested to ensure proper interpretation of the test result. This testing of the family member may not be covered by health insurance.

These five features of genetic testing pose additional challenges to the health care financing and delivery system. Given that genetic testing is growing and genetics is becoming more important in clinical practice and public health, the system should be better equipped to provide and pay for genetic services and followup care appropriately, effectively, and in a coordinated manner.

A Discussion of Genetic/Genomic Services

Genetic/genomic services are types of health services provided by laboratories and various health providers, including primary care physicians, medical geneticists, pathologists, genetic counselors, and genetic nurses. They include laboratory services that involve the provision of tests using genetic/genomic technologies, interpretation of results, and oversight of the test's performance. Other genetic/genomic services include identification or diagnosis of individuals and families at risk for a disorder with a genetic component or who could benefit from pharmacogenomic testing. These services also include provision of support and genetic counseling to patients; facilitation of genetic/genomic testing; assistance with the interpretation of test results; explanation of germline, inherited, and acquired disorders; analysis of inheritance patterns; review of the potential options for intervention; and management of clinical treatment.

Health Provider Terminology

Several terms for health providers are used throughout this report. The following descriptions are provided to clarify how the terms are used:

Health providers refers broadly to all practicing physicians and nonphysician health providers.

Nonphysician health providers refers to those practicing health providers who do not have an MD or DO degree but who are qualified to provide health care services to patients.

Genetics health providers refers to a subset of health providers who have specialized training in genetics. The term includes but is not limited to medical geneticists, PhD geneticists, certified genetic counselors, and genetics nurses.

Genetic counseling providers refers to physicians and nonphysician health providers who provide genetic counseling services to patients.

Test Characteristics

Analytical validity refers to how well a test performs in the laboratory—that is, how well the test measures the property or characteristic it is intended to measure. In other words, does the test do what its makers claim it does? If so, it must produce the same results repeatedly and in different laboratories (given the same set of procedures).

Clinical validity refers to the accuracy with which a test predicts the presence or absence of a clinical condition or predisposition. Initially, the test has to be conducted on individuals who are known to have the condition (as well as those who do not) to determine its success rate.

Clinical utility refers to the usefulness of the test and the value of information to the person being tested. If a test has utility, it means that the results—positive or negative—provide information that is of value to the person being tested because he or she can use that information to seek an effective treatment or preventive strategy. Even if no interventions are available to treat or prevent disease, there may be benefits associated with knowledge of a result.

Source: Secretary's Advisory Committee on Genetic Testing, Enhancing the Oversight of Genetic Tests, June 2000. http://www4.od.nih.gov/oba/sacgt/reports/oversight_report.pdf [November 18, 2005].

A Discussion of Genetic/Genomic Tests and Technologies

Genetic/genomic technologies are processes or methods used to analyze human DNA, RNA, genes, chromosomes, proteins, or metabolites that detect mutations, chromosomal changes, karyotypes, phenotypes, and/or expression pattern variation. Genetic/genomic technologies are applied to tests for germline, inherited, and/or acquired variations in the genome, transcriptome, and proteome. Genetic tests generally focus on testing one or a few genes, whereas genomic tests assess larger numbers of genes and sequences up to the context of the entire genome. Throughout this report, use of the terms "genetic test," "genetic technology," or some variation thereof implies inclusivity of all genetic and genomic technologies.

Historically, genetic tests have been used to identify germline or heritable variations in an individual's genome, whether analyzing DNA, RNA, or proteins. Currently, the term "genetic test" is used more broadly to refer to any test performed using molecular biology methods to test DNA or RNA, including germline, heritable, and acquired somatic variations. As we advance toward genomic medicine, with acquired somatic variations evaluated in the context of an individual's entire genome variations, the definition of a genetic test will become even broader. Therefore, although this report focuses on genetic tests and services with a narrower definition, it is SACGHS's intention that lessons learned from genetic tests and services be applied to future innovations in clinical care developed using genetic/genomic technologies involving germline, inherited, and acquired alterations. However, because tests for germline heritable variations have more implications for all blood relatives of an individual patient compared with somatic acquired variations, in some contexts, including but not limited to science policy, testing oversight, and ethical contexts, the narrower definition of a genetic test as a test for a germline and/or heritable alteration, and not for somatic variants, should be used.

Genetic/genomic tests can be used to diagnose a disease, predict future disease, predict risk or susceptibility to disease, direct clinical management, identify carriers of genetic mutations, and establish prenatal or clinical diagnosis or prognosis in individuals, families, or populations. Genetic/genomic tests may be used, for example in preimplantation diagnosis and newborn screening.

Predictive testing determines the probability that a healthy individual might develop a certain disease in the future. For example, a test for the Huntington disease gene may be used to predict future disease in individuals with a family history of the disease. A predisposition test such as *BRCA1/2* testing for breast cancer may indicate an increased risk for disease but does not definitively predict onset.

Pharmacogenetic/pharmacogenomic tests are used to determine the likelihood of a person being responsive to a particular drug and/or having an adverse event. For instance, pharmacogenetic/pharmacogenomic testing for the presence of a thiopurine S-methyltransferase (*TPMT*) mutation can determine whether patients with acute lymphoblastic leukemia will have a potentially fatal response to the standard dose of the chemotherapeutic agent 6-mercaptopurine.

The specific examples noted above are for germline and heritable mutations, but the same genetic/genomic testing methods can be used to detect acquired mutations. Detection of *RAS* mutations in stool for colorectal cancer is an example of a test for an acquired mutation present in the colon cancer cells. Testing for noninheritable variations generally does not raise as many ethical, legal, and social issues for patients and family members as genetic tests for heritable mutations.

Genetic/genomic technologies used for nonmedical purposes such as forensic identification or establishing paternity or familial relationships are not considered in this report.

Coverage

Every public program and private health insurance plan[10] outlines the scope of services it will cover, the circumstances under which the payer will reimburse, and any cost-sharing components (e.g., deductibles, copayments) or coverage limits. Because coverage decisions are made on a plan-by-plan basis, patient access to genetic tests and services vary by health insurance plan.

Coverage Decisions

Overview of Coverage Decisionmaking. In general, coverage decisionmaking is the process by which health insurance plans and purchasers determine which services they will cover as well as the circumstances under which they will reimburse providers for the service or procedure (e.g., strong family history, age). These determinations are dependent on many factors, including the quality of existing clinical evidence, consumer or purchaser demand, and State and Federal laws (e.g., mandated benefits or Medicare/Medicaid laws).

As new information becomes available and as new tests or services are developed, modifications may be made to existing coverage policies, or new policies may be issued granting or excluding coverage for a particular service. Member or employer requests, new State mandates, or fiscal considerations can prompt or influence such decisions. Benefit policies also are strongly influenced by the actions of CMS and large private health plans.

Coverage decisionmaking also occurs when a claim for reimbursement or a request for preauthorization is submitted. If the service is covered or at least not explicitly excluded from coverage, a determination is made about whether the circumstances of the individual plan member meet the coverage criteria. If they do, then the service usually is covered. If the service is not specifically addressed by current coverage policies or the patient's particular circumstances are not discussed in the existing coverage criteria, further review may be necessary to adjudicate the claim.

Private Plan Coverage of Genetic Tests and Services. Because the coverage policies of private health insurance plans are considered proprietary, public access to policy information is limited.[11] As a result, it is difficult to assess which genetic tests and services tend to be covered in private plans and which ones are not covered as well as the circumstances for and extent of coverage. Of the few coverage policies that are publicly available, most cover genetic testing for chromosomal abnormalities, prenatal and neonatal diagnosis, and, in some cases, preimplantation genetic diagnosis in certain situations (e.g., advanced maternal age, suspected fetal anomaly, or history of miscarriage or developmental problems in prior

[10] A broad overview of health insurance and health care financing in the United States is provided in Appendix A.

[11] Federal and State laws require health insurance plans to provide plan members with information about covered services and explanations of how coverage decisions are made and how they can be appealed. If a claim is denied, health insurance plans also are required to provide plan members and their providers with a reason for the denial, citing any policy or criteria on which the decision is based.

pregnancies). Also, these publicly available policies usually cover diagnostic genetic testing for rare diseases under general policies for genetic tests. Some health plans have policies for specific conditions, such as hereditary cancer testing, cystic fibrosis, Tay-Sachs disease, and hereditary hemochromatosis. An example of an explicit noncoverage policy is the exclusion of genetic testing for Alzheimer's disease. The stated reason for exclusion is that the test is considered experimental.

The following coverage criteria were cited in publicly available plans:

- The patient has current signs and/or symptoms (i.e., the test is being used for diagnostic purposes).
- Conventional diagnostic procedures are inconclusive.
- The patient has risk factors or a particular family history that indicate a genetic cause.
- The patient meets defined criteria that place them at high genetic risk for the condition.
- The test is not considered experimental or investigational.
- The test is performed by a CLIA-certified laboratory.
- The test result will directly influence the disease treatment management of the covered member.
- In some cases, testing is accompanied by pretest and posttest counseling.

Generally, genetic testing is not covered for:

- Population screening without a personal or family history, with the exception of newborn screening and preconception or prenatal carrier screening for certain conditions, such as cystic fibrosis, Tay-Sachs disease, sickle cell disease, and other hemoglobinopathies
- Informational purposes
- Minors for adult-onset conditions
- A relative of a plan member who is not also a plan member unless (1) the genetic test results are necessary for the medical care of the plan member and (2) the relative can provide evidence of coverage denial from his or her health insurance plan[12]

Evidence-Based Coverage Decisions. Health care cost constraints, demand for improved health outcomes, a greater emphasis on quality, and the introduction of new technologies and procedures available for clinical use all are driving health care payers to reassess how they make decisions about which tests and services to cover and the conditions under which they will reimburse them. Health insurance plans have emphasized evidence-based coverage decisionmaking as an effective way to determine which technologies and services are appropriate to cover.

Role of Technology Assessments. Technology assessments have an important role in coverage decisionmaking. They involve evaluating existing evidence to ascertain whether the test or service has been found to be safe, effective, and appropriate for coverage and reimbursement. Some health insurance plans and purchasers have established comprehensive, internal processes for conducting these technology assessments; others with fewer resources perform less formal reviews or purchase technology assessments conducted by other technology assessment groups such as Blue Cross Blue Shield Association, ECRI, or HAYES, Inc. These technology assessments are critical to ensuring that coverage decisions are based on sound evidence.

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[12] To our limited knowledge, Aetna is the only health insurance plan that will cover genetic testing for relatives under these circumstances.

Blue Cross Blue Shield's Technology Evaluation Center (TEC) systematically evaluates the clinical effectiveness and appropriateness of new technologies (but does not issue coverage decisions). Their technology assessments and review criteria have been used industry-wide to make coverage decisions.

TEC uses the following criteria for assessing new technologies:

- When applicable, the technology must have final approval from the appropriate governmental regulatory bodies.[13,14]
- The scientific evidence must permit conclusions concerning the effect of the technology on health outcomes. Specifically:
  - Evidence should be derived from well-designed, well-conducted investigations that are published in peer-reviewed journals. The quality of the body of studies and the consistency of results are considered in evaluating the evidence.
  - The evidence should demonstrate that the technology can measure or alter the physiological changes related to disease, injury, illness, or condition. In addition, there should be evidence or a convincing argument based on established medical facts that such measurement or alteration affects health outcomes.
  - Opinions and evaluations by national medical associations, expert panels, or other technology evaluation bodies are considered in the context of the quality of the supporting evidence and rationale.
- The technology must improve the net health outcome (i.e., the technology's beneficial effects on health outcomes should outweigh any harmful effects).
- The technology must be at least as beneficial as any established alternative.
- The improvement must be attainable outside the investigational setting when used under the usual conditions of medical practice.[15]

Evidence-Based Coverage of Genetic Tests. For genetic tests, evidence-based coverage decisionmaking involves assessing existing data on a test's analytical and clinical validity and clinical utility as it relates to the populations served by the health insurance plan (e.g., for Medicare, data on individuals age 65 and older should be available). Coverage decisionmaking also involves consideration of other factors such as the test's cost[16] and patient preference.

Some genetic tests that currently are available clinically are supported by few or no clinical data, and thus, may not provide useful, accurate or interpretable results.[17] The historic focus on basic research instead of on translational research has contributed to an insufficient accumulation of clinical data to satisfy evidence

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[13] TEC will consider indications for which the technology has not been formally approved (i.e., off-label use).

[14] Laboratory-developed genetic tests, which account for the majority of genetic tests, are not subject to premarket review by the Food and Drug Administration (FDA). Genetic tests that are packaged and sold as kits, on the other hand, are subject to FDA review.

[15] Technology Evaluation Center Criteria. http://www.bluecares.com/tec/teccriteria.html [November 18, 2005].

[16] Medicare expressly does not consider the cost or cost-effectiveness of a technology or service when making coverage decisions.

[17] Higashi MK and Veenstra DL. Managed care in the genomics era: Assessing the cost effectiveness of genetic tests. *Am J Manag Care* 2003. 9(7): 493-500.

Considerations in Making Coverage Decisions for New Technologies

- Is it FDA approved, cleared, or not subject to FDA review?
- Do clinical trials demonstrate medical effectiveness and improved health outcomes?
- Are there practice guidelines that recommend its use?
- What do the experts and professional organizations say about it?
- Is it experimental?
- How much does it cost?
- Is it cost-effective?
- Are there any administrative, social, legal, or political factors that should be considered?
- Is there demand for its coverage?
- Do other health care payers already cover it?
- Is a CPT code available for billing?

standards for genetic test coverage decisions.[18] Some observers suggest that lack of regulation by the Food and Drug Administration of "home brew" laboratory tests (which represent the majority of tests) creates less incentive for genetic test developers to amass clinical validity and utility data that could be used to support evidence-based decisions, thus contributing to an insufficient evidence base for genetic tests.[19]

The novelty and predictive nature of genetic tests present additional challenges to the standards traditionally used to assess the evidence base for coverage decisionmaking. These challenges are described below.

Informational Utility. As demonstrated by the TEC criteria above, health insurance plans increasingly require technologies and services to demonstrate improved health outcomes. Meeting this criterion can be challenging for most diagnostic tests[20] and especially for genetic tests for conditions that lack therapeutic and preventive options. Even though there may be no therapeutic options associated with a genetic test result, many people may find this information to be useful for purposes of family and estate planning and preparing for long-term care needs.

Tests lacking therapeutic options raise the question of whether the information provided is significant and meaningful enough in a health care context to warrant coverage by payers. In addition, they may raise broader questions about the appropriateness of expending finite health care resources on a test that lacks treatment options and the extent to which health insurance coverage should extend beyond the role of health maintenance and promotion. These questions will need to be explored in any discussion of genetic testing, since many presymptomatic genetic tests will become available in the absence of effective clinical interventions, either preventive or therapeutic.

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[18] The National Institutes of Health's Roadmap Initiative aims to increase focus on translational research that "transforms our new scientific knowledge into tangible benefits for people" (http://nihroadmap.nih.gov/overview.asp) [November 18, 2005].
[19] Public comments to SACGHS on draft Report on Coverage and Reimbursement of Genetic Tests and Services. June 2005.
[20] "Impacts of diagnostic tests on health outcomes often are confounded by variable effects of treatments or interventions initiated following diagnostic use. Collecting direct evidence to substantiate a link between diagnostic use and resulting health outcomes is, at least, challenging and is sometimes infeasible." (Prepared for AdvaMed by the Lewin Group. The Value of Diagnostics: Innovation, Adoption and Diffusion into Health Care. July 2005. http://www.advamed.org/publicdocs/thevalueofdiagnostics.pdf [November 18, 2005]).

Recouping Short-Run Costs. Because plan members change plans at fairly frequent intervals, private health insurance coverage for preventive services can be difficult to rationalize from an economic standpoint since the cost savings may not accrue to the plan that paid for the service. This challenge is of particular concern to health insurance plans when coverage for the test is not widespread throughout the health insurance market. Also, this concern is more prevalent when the test is first introduced into clinical practice and it is uncertain to what extent the market will cover it. Health insurance plans covering such tests and services need to calculate whether it makes economic sense to provide genetic tests and followup care even though they do not stand to reap future savings (in effect, they are sparing a future health insurance plan from the downstream costs associated with treating the illness at a later date).[21,22]

Amassing Sufficient Evidence for Translation of Genetic Tests Into Clinical Use. Many genetic tests may be excluded from coverage due to the "experimental" nature of the technology or service and insufficient evidence. Like many tests, determining when genetic tests have moved beyond the research phase and thus are no longer experimental, can be difficult because their transition from research to clinical use is not clear-cut. This is a particular problem for genetic tests for rare diseases. Establishing clinical validity and amassing sufficient data to achieve statistically significant results that satisfy evidence standards are especially difficult for rare diseases due to their low prevalence. In addition, scientific interest in any given rare disease also is a factor affecting data collection (and access to research testing). A specific rare disease may be studied by only one investigator in the country or world. Insufficient funding for the study of rare diseases also can impede the amassing of adequate data.

What Is Cost-effectiveness Analysis?

Cost-effectiveness analysis (CEA) is a method of estimating the costs and health outcomes of an intervention relative to some alternative (e.g., no intervention, alternate intervention, administering intervention less frequently or to fewer individuals). The cost-effectiveness of an intervention compared to its alternative is often reported as the difference in cost per quality-adjusted life-year (QALY) gained.

CEAs can be used to inform decisions about health care resource allocation. Individuals responsible for making resource allocation decisions must decide whether the improvement in effectiveness is worth the additional cost.

Source: Gold MR et al. Cost-effectiveness in Health and Medicine. New York: Oxford University Press, 1996.

Role of Cost-Effectiveness Data. Cost-effectiveness data, when available and appropriate, are one piece of information among many that health insurance plans can use in making evidence-based coverage decisions. Health insurance plans' interest in economic data is especially heightened for expensive, novel tests, when downstream followup or treatment costs are substantial, or when several similar products are available. Genetic tests have not yet had a significant enough impact on health plan budgets to be a priority for economic evaluations; however, as utilization of pharmacogenetic and genetic tests for common diseases

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[21] Phillips KA et al. Genetic testing and pharmacogenomics: Issues for determining the impact to health delivery and costs. *Am J Manag Care* 2004. 10(7):425-32.

[22] Williams MS. Can genomics deliver the promise of improved outcomes and reduced costs? Background and recommendations for health insurers. *Disease Management and Health Outcomes* 2003. 11(5):277-90.

becomes more widespread and drives consumption[23] of more resources, interest in cost-effectiveness data for genetic tests is likely to intensify.

Cost-effectiveness data can be particularly useful in supporting coverage decisions if the test obviates the need for other tests or treatment, helps avoid ineffective interventions, or allows for less intensive (and presumably less costly) treatment due to earlier detection.[24] Preventive services are covered by most managed care organizations because they are particularly valued by healthy potential plan members and their per-person cost is low relative to per-person disease treatment costs.[25]

Several characteristics are important to consider when conducting economic evaluations of genetic tests:[26]

- Genotype-phenotype association (clinical validity). There should be sufficient evidence of an association between the gene variant and clinically relevant phenotypes.
- Prevalence of the gene variant(s). The prevalence of a gene variant(s) will have a significant impact on the cost-effectiveness of a genetic test and should be described for the patient population of interest
- Outcome characteristics. The severity of the outcomes associated with the gene variant should be considered, potentially including patient quality of life, mortality, and economic costs.
- Intervention for the variant group (clinical utility). An intervention guided by the genetic test results should be specified, and the risk reduction for that intervention should be quantified, preferably as measured by the attributable risk reduction.
- Test characteristics. The test sensitivity and specificity (analytical validity) should be considered as well as direct and indirect costs associated with the test, including genetic counseling costs and induced costs such as additional provider visits.[27,28]

Thus far, relatively few cost-effectiveness analyses have been conducted for genetic tests.[29] Of those that have been performed, results have been mixed. Such results often have been sensitive to the prevalence of the genetic mutation in the population, disease characteristics, severity of outcomes, treatment costs and length, and accuracy of the genetic test. The low number of favorable cost analyses and limited data available to conduct such studies may be affecting the willingness of the health plans to consider coverage of genetic tests and services.

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[23] Direct-to-consumer advertising and insurance coverage could increase uptake and thereby increase the economic impact of genetic testing.

[24] Williams MS. Can genomics deliver the promise of improved outcomes and reduced costs? Background and recommendations for health insurers. *Disease Management and Health Outcomes* 2003. 11(5):277-90.

[25] The low per-person cost of preventive services may be attributable to high utilization and low equipment and time costs. In some cases, however, it may be due to low reimbursement rates for these services.

[26] These factors are also important in assessing the clinical utility of the test.

[27] Higashi MK and Veenstra DL. Managed care in the genomics era: Assessing the cost effectiveness of genetic tests. American Journal of Managed Care 2003. 9(7): 493-500.

[28] Patients seeking genetic services frequently have medical problems that, depending on the test results and nature of the disease and test (e.g., presymptomatic, diagnostic), may require additional testing, more frequent periodic screening, or long-term treatment whose costs can be substantial. Thus, when costs are taken into consideration in coverage decisionmaking, it is important to consider not only the immediate costs of the genetic services (e.g., cost of the test and counseling) but also the costs associated with any followup care appropriate for the patient's circumstances. Not including these costs could result in a poorly informed coverage decision.

[29] Phillips KA and Van Bebber SL. A systematic review of cost-effectiveness analyses of pharmacogenomics interventions. *Pharmacogenomics* 2004. 5(8):1139-49.

Considering cost-effectiveness data as part of coverage decisionmaking has been controversial, but some are beginning to acknowledge its value. For example, the U.S. Preventive Services Task Force has begun reviewing published cost-effectiveness studies as part of its development of recommendations for clinical preventive services, although these studies do not influence its recommendations. In its report to Congress, the Medicare Payment Advisory Commission identifies several ways in which Medicare could begin to consider cost-effectiveness, including the collection of cost-effectiveness information from manufacturers and providers in the coverage process.[30] Appropriately incorporating cost considerations in coverage decisionmaking requires answering hard questions about how cost-effectiveness data ought to affect decisions and what threshold should be used for determining whether a service is cost-effective.

Although cost-effectiveness data, in conjunction with other information, can be useful to consider when making coverage decisions, it is important to acknowledge the inherent limitations of the utility of cost evaluations and that there may be situations where they are either inappropriate or infeasible. For example, genetic testing for rare diseases, which are characterized by their low prevalence, likely would never be found to be cost-effective. However, such genetic tests clearly have value, as demonstrated by States' inclusion of rare disease tests in their newborn screening programs.

Recommendation 1

The Secretary should task an appropriate group to develop a set of principles to guide coverage decisionmaking for genetic tests and services. The guiding principles should address the issues identified in this report, including economic evaluation/cost-effectiveness, prevention, rare disease tests, therapeutic benefit, and informational utility. The group also should assess the type, quality, and quantity of existing evidence for specific genetic tests to determine whether the evidence is adequate to establish a test's analytical validity, clinical validity, and clinical utility. If not, the group should identify any evidentiary gaps.

This group should consist of experts from both the public (i.e., HHS agencies) and private sectors and make use of resources or models from both sectors. A work group organized by the Centers for Disease Control and Prevention, called the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Work Group (see box on page 24), is an example of such a group. It is made up of a diverse range of experts from both sectors and is performing related work and, thus, could be tasked to develop these principles.

In addition, a mechanism should be established to promote and fund studies to address evidentiary gaps identified by the group.

Role of Consumer Demand. In addition to evidence-based coverage decisionmaking, consumer demand has been shown to influence coverage decisions. For example, some health insurance plans granted coverage for high-dose chemotherapy plus autologous bone marrow transplant under public pressure, even

[30] MedPAC. Report to Congress: Issues in a Modernized Medicare Program, June 2005. http://www.medpac.gov/publications/congressional_reports/June05_ch8.pdf [November 18, 2005].

ACCE/EGAPP Projects: HHS Efforts to Review the Existing Evidence Base for Genetic Tests Entering Clinical Practice

Sponsored by the Centers for Disease Control and Prevention (CDC), the ACCE Project developed and tested a model system for identifying, synthesizing and disseminating existing data on DNA-based tests and for identifying gaps in knowledge. The ACCE process got its name from the four components of evaluation proposed by the Task Force on Genetic Testing and the Secretary's Advisory Committee on Genetic Testing – analytical validity, clinical validity, clinical utility, and ethical, legal, and social implications. The ACCE reviews provided summary information to support decision making but did not present conclusions or recommendations.

The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Project is a 3-year pilot project that aims to build on experience from the ACCE Project and prior recommendations for action, and to collaborate with stakeholders, other agencies and existing evidence-based processes (e.g., U.S. Preventive Services Task Force) in order to establish and evaluate a systematic mechanism for pre- and post-market assessment of genomic applications. Health insurance payers and purchasers are among the project's intended audience. The EGAPP Project will establish an independent Working Group with roles that include: selecting and prioritizing topics for review; overseeing expert and peer review of reports; developing conclusions and recommendations based on the evidence; and considering the needs and strategies for post-implementation monitoring and data collection.

though the evidence to support coverage was weak or nonexistent.[31] With regard to genetic tests, direct-to-consumer marketing may be having an impact on consumer demand, especially given greater access to health information through the Internet and other media.[32]

Evidence Base for Genetic Counseling Services. Like genetic tests, measuring the clinical effectiveness of genetic counseling services is challenging due to the difficulty in demonstrating a causal association between counseling and health outcomes, especially given the emphasis on nondirectiveness. Researchers have used several measures to assess the value and effectiveness of genetic counseling services. Various studies have shown increased knowledge, lower costs as a result of more appropriate use of genetic tests, and higher rates of risk identification as some of the positive outcomes of genetic counseling services.[33] Genetic counseling services provided by nonphysician providers also can lead to cost containment since nonphysician providers typically charge 20 to 50 percent less than physicians.[34]

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[31] Even though there was a paucity of clinical evidence of efficacy, more than 41,000 patients underwent high-dose chemotherapy plus autologous bone marrow transplant (ABMT) for breast cancer in the 1990s. Intense political lobbying, the threat of litigation (exacerbated by a court decision in California that awarded $89 million to the family of a woman who was denied coverage for ABMT and eventually died from breast cancer), and several State and Federal mandates caused many health plans to cover the treatment. (Mello MM and Brennan TA. The controversy over high-dose chemotherapy with autologous bone marrow transplant for breast cancer. *Health Affairs* 2001. 20(5):101-117)

[32] SACGHS wrote a letter to the Secretary in December 2004 requesting that HHS agencies conduct an analysis of the public health impact of direct-to-consumer advertising of genetic tests.

[33] A more detailed discussion of the value and effectiveness of genetic counseling services is provided in Appendix B.

[34] Gibons A. Employer-based coverage of genetic counseling services. *Benefits Quarterly* 2004, p.48-68.

Recommendation 2

Although standardization of coverage decisions using the best scientific evidence across public and private payers is ideal (see Recommendation 1), private health insurance plans are encouraged to make their own coverage determinations about genetic tests and services relative to the populations they serve. The group described in Recommendation 1 should make available the scientific evidence needed to make these decisions.

Influence of Medicare on Private Plans. Because Medicare is the largest provider of health insurance in the United States, its coverage decisions are closely monitored by private health insurance plans. When Medicare decides to cover a test or service, the private market very often follows these decisions. It is not clear whether this reliance on Medicare's decisionmaking is appropriate with respect to genetic tests. Because genetic tests often are used for preventive, reproductive, or life planning purposes and because most hereditary diseases manifest prior to age 65, the utility of many genetic tests and services in the Medicare population is not straightforward. Since the patient population is a relevant factor in coverage decisionmaking, it may not be appropriate for private health insurance plans to follow Medicare's lead in making coverage decisions for predictive and predispositional genetic tests and services. Perhaps in realizing that Medicare policy may not be the most appropriate model, some health plans have not waited for Medicare's assessment prior to covering a genetic test or service.

Medicare Coverage

Although Medicare faces many of the same challenges as private health insurance plans, several are unique to or experienced differently by the Government program, including how coverage decisions are made and which services can be considered for coverage.

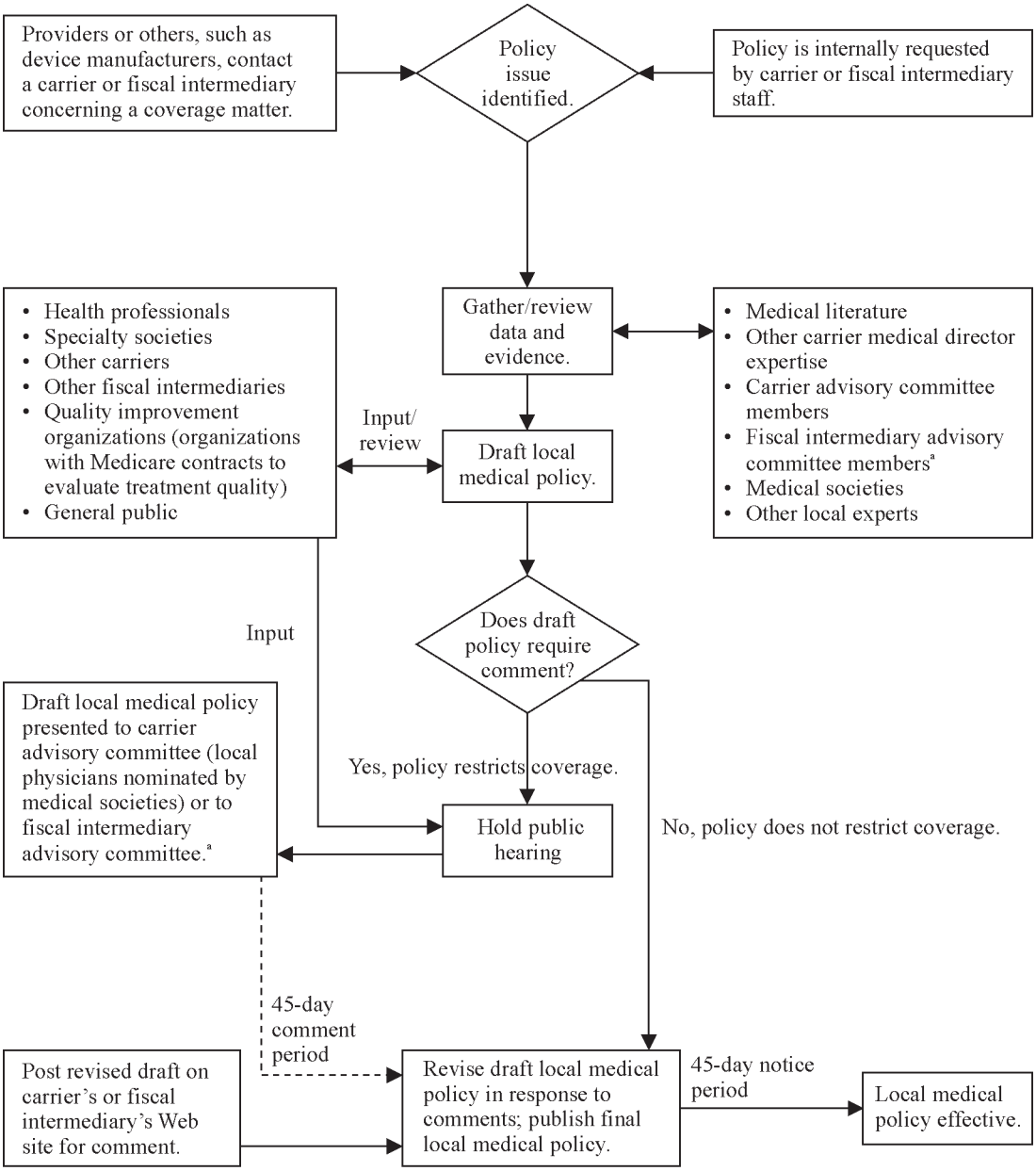
Role of Congress. When Medicare was established in 1965, Congress broadly outlined the scope of benefits that were covered by the program (see page A-5 for benefit categories). Congress continues to have a role in defining the scope of benefits, as evidenced by the recent addition of prescription drug benefits.

Role of the Centers for Medicare & Medicaid Services. The Centers for Medicare & Medicaid Services (CMS) is responsible for interpreting and implementing Medicare law. More specifically, the agency determines whether specific services fall within the congressionally defined benefit categories and decides whether to add a service to the scope of benefits. CMS also has a role in establishing and implementing policies that guide coverage decisions.

Medicare Coverage Advisory Committee. CMS may seek the assistance of the Medicare Coverage Advisory Committee (MCAC) when a service to be considered for coverage is the subject of significant scientific, medical, or public controversy; has the potential to have a major impact on the health of beneficiaries and/or the Medicare program itself; or raises important social, legal, or ethical issues. MCAC is responsible for evaluating and advising CMS on the effectiveness and appropriateness of medical services or items.

Local Coverage Determinations. Medicare coverage decisions are made at both the local and national levels, although the majority of Medicare coverage policy decisions are made by the 23 local Medicare administrative contractors. Local policies apply only to the area that the contractor serves and must adhere to

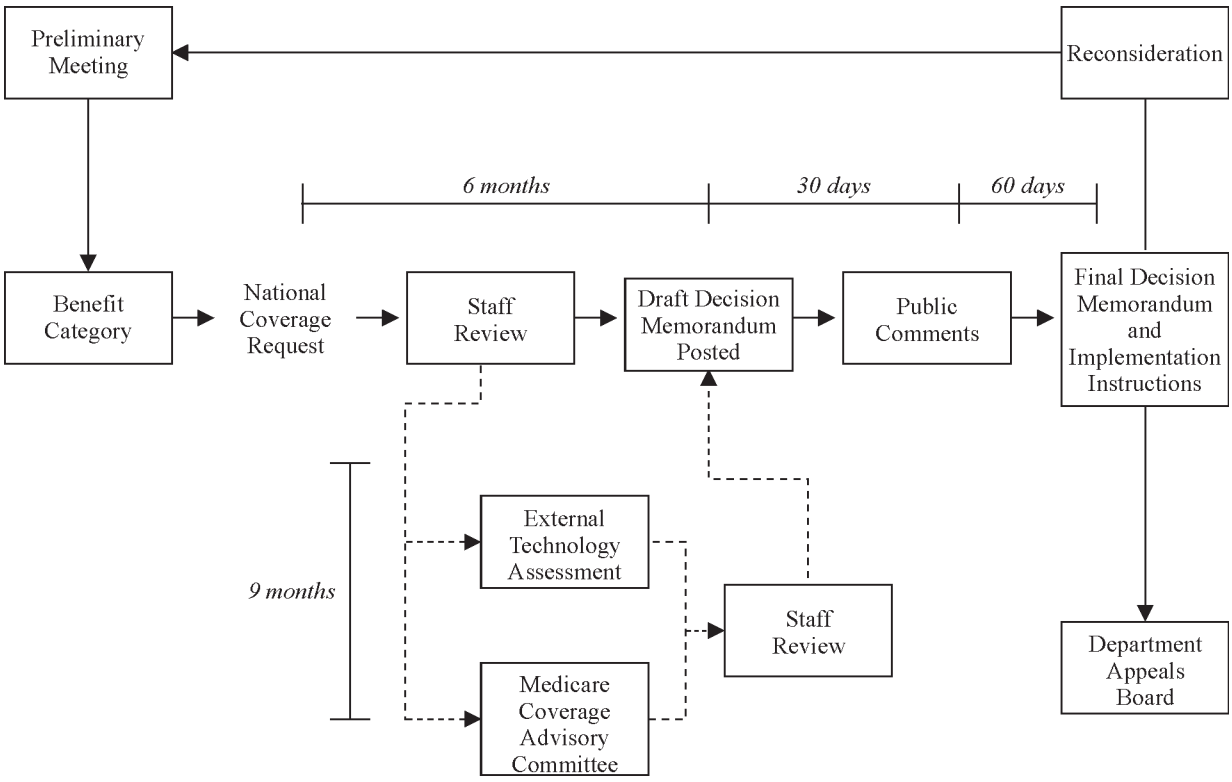
Medicare Local Coverage Review Process



Source: Government Accounting Office. Medicare: Divided Authority on Policies for Coverage of Procedures and Devices Results in Equalities, April 2003. http://www.gao.gov/new.items/d03175.pdf.

[a] Fiscal Intermediaries may have advisory committees, but CMS does not require them to do so.

Medicare National Coverage Process



Source: Sean Tunis, Medicare Coverage and Genetic Testing. Presentation to SACGHS on March 1, 2004. http://www4.od.nih.gov/oba/SACGHS/meetings/March2004/Tunis.ppt.

national statutes and regulations. This approach allows Medicare to be responsive to geographic variations in clinical practice and beneficiary needs and to extend coverage to new tests and services more rapidly than the national coverage process. Local coverage determinations (LCDs) also can help manufacturers collect the necessary evidence to support a national coverage determination (NCD).

National Coverage Determinations. NCDs are made at the Federal level and apply to all beneficiaries and local administrative contractors. National coverage review processes can be initiated internally within CMS or through a formal request from a member of the public. A review process may be initiated internally for many reasons, including if:

- There are conflicting LCDs.
- The service represents a significant medical advance with no comparable covered service.
- There is a question about the service's clinical effectiveness or about the appropriate administration of the service.
- The service is covered but widely held to be obsolete or ineffective.
- There are concerns about overutilization or underutilization.[35]

[35] 64 FR 22619. Procedures for Making National Coverage Decisions.

Advantages and Disadvantages of Local Medicare Coverage Processes

Advantages	Disadvantages
• Allows Medicare the flexibility to respond to unique regional health care needs and challenges • Allows contractors the flexibility to manage utilization differences, ensure appropriate billing, and address instances of abusive billing swiftly • Provides greater opportunities for provider, manufacturer and other stakeholder input on coverage policies • Offers patients and practitioners access to new technologies that may not otherwise exist if all Medicare coverage decisions were made at the national level • Provides opportunities to accrue evidence based on the safety, effectiveness, and costs associated with technologies • Offers opportunities to characterize the circumstances of use for technologies • Provides early revenue generation opportunities for manufacturers to support ongoing product development that may not otherwise exist	• Significant potential for establishing variable and sometimes inappropriate coverage policies that affect patient access and quality • Significant variations in the criteria and methods used to make coverage decisions • Less than 50% of the LCDs made by the 46 Medicare carriers and fiscal intermediaries cited peer-reviewed clinical evidence[1] • Differing availability of resources to conduct or purchase technology assessment services • Creation of coverage policies that limit or expand coverage of certain tests beyond the number per patient recommended for clinical practice (can be precipitated by a spike in claims) • Decentralized system that often results in redundant coverage efforts • Frequent criticism for untimely and uncoordinated mechanisms to raise eligible coverage policies for national consideration

[1] Foote SB et al. Resolving the tug-of-war between Medicare's national and local coverage. *Health Affairs* 2004. 23(4):108-23.

Source: Prepared for AdvaMed by the Lewin Group. The Value of Diagnostics: Innovation, Adoption and Diffusion into Health Care. July 2005. http://www.advamed.org/publicdocs/thevalueofdiagnostics.pdf [December 5, 2005].

Impact of National-Local Decisionmaking Process. Although the current combination of national and local systems used by Medicare for making coverage decisions is intended to maximize regional flexibility, this system can create impediments to securing coverage in certain circumstances. Different local coverage policies can lead to inconsistencies in coverage from one region to another. Entering into the national coverage review process is not without risk, however. The process can result in a noncoverage or a limited coverage decision. In such cases, the NCD preempts any existing LCDs for the technology or service and prevents implementation of future LCDs. Also, Medicare decisions are closely followed by private health plans, so noncoverage decisions also can affect coverage by private health plans. Furthermore, although it is possible to appeal a noncoverage decision, it is an extremely lengthy process with multiple requirements.

BRCA 1/2 Testing: An Example of Variation in National v. Local Coverage Decisions

Most local Medicare administrative contractors do not cover predictive *BRCA1/2* testing because they consider it to be a screening test, which CMS has interpreted not to be a statutory benefit. However, a few local Medicare administrative contractors have decided to allow coverage of *BRCA* testing performed in the absence of signs, symptoms, or personal history of the disease. The result is that Medicare coverage of the *BRCA1/2* genetic test varies depending on where in the United States the beneficiary lives.

Recommendation 3

The Secretary should encourage CMS to move forward with the development of a plan to evaluate new local coverage decisions to determine which should be adopted nationally and to what extent greater consistency in Medicare coverage policy can be achieved (such a plan is mandated in Section 731 of the Medicare Prescription Drug, Improvement, and Modernization Act of 2003). As part of its implementation of Section 731, CMS should consider a mechanism that would automatically initiate a national coverage review process for any test or service approved for coverage by a certain number of local Medicare administrative contractors.

FDA Approval Requirement. To be considered for Medicare coverage, tests under the purview of the Food and Drug Administration (FDA) must be FDA-approved.[36,37] In the case of laboratory-developed genetic tests, which are not subject to FDA premarket review (but for which FDA approval can be sought), this criterion is not applicable. Genetic test kits that are packaged and sold commercially, on the other hand, are subject to FDA review and must be proven to be safe and effective for clinical use before CMS will consider coverage.

Reasonable and Necessary Requirement. Tests and services covered by Medicare also must be shown to be "reasonable and necessary" for the diagnosis and treatment of illness or injury or to improve the functioning of a malformed body member. Although there is no statutory or regulatory definition of reasonable and necessary, CMS has interpreted the phrase to require that the item or service should, at a minimum, improve net health outcome for Medicare beneficiaries. To reach a conclusion about whether an item or service is reasonable and necessary, CMS uses standard principles of evidence-based medicine, which require a thorough evaluation of relevant clinical evidence to determine whether the evidence is of sufficient quality. The assessment of clinical evidence is divided into three stages:

1. Quality of the individual studies
2. Relevance of findings from individual studies to the Medicare population
3. Overarching conclusions that can be drawn from the body of the evidence on the direction and magnitude of the technology's risks and benefits

Possible Outcomes of Coverage Decisionmaking Process. The following are possible outcomes of CMS's coverage decisionmaking process:

1. A national coverage decision is issued with limitations on coverage.
2. A national coverage decision is issued with no limits on coverage.
3. A national noncoverage decision is issued precluding local administrative contractors from making payment.
4. No national decision is issued, leaving coverage to the discretion of the local contractor.

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[36] CMS. What is the difference between FDA and CMS review? http://questions.cms.hhs.gov/cgi-bin/cmshhs.cfg/php/enduser/std_adp.php?p_faqid=2656&p_created=1079987789&p_sid=BvLmBNYh&p_lva=&p_sp=cF9zcmNoPSZwX3NvcnRfYnk9JnBfZ3JpZHNvcnQ9JnBfcm93X2NudD0xNDcwJnBfcHJvZHM9JnBfY2F0cz0mcF9wdj0mcF9jdj0mcF9zZWFyY2hfdHlwZT1hbnN3ZX [January 4, 2006].

[37] Certain non-experimental/investigational devices used in clinical trials conducted under Investigational Device Exemptions can be considered for Medicare coverage (Prepared for AdvaMed by the Lewin Group. The Value of Diagnostics: Innovation, Adoption and Diffusion into Health Care. July 2005. http://www.advamed.org/publicdocs/thevalueofdiagnostics.pdf [November 18, 2005]).

Medicare Coverage of Genetic Tests. Of the approximately 274 national coverage decisions issued by CMS, only one relates to genetic tests and services—cytogenetic analyses for monitoring acute leukemia, myelodysplasia, and congenital abnormalities. Although a few local Medicare coverage policies have been developed for HER-2/*neu* and *BRCA* testing, most local administrative contractors do not cover them because they consider it to be a screening test, which CMS has interpreted not to be a statutory benefit.

Screening Exclusion. In order for Medicare to cover a service or technology, it must fall within one of the statutorily authorized benefit categories. Items and services that do not fit within one of the benefit categories prescribed by Congress are not reimbursable.

Relevance of Genetic Tests to the Medicare Population

Although predispositional genetic tests are of primary benefit to younger individuals, there are many genetic tests that are relevant to the elderly Medicare population. For example, frontotemporal dementia with Parkinsonism linked to chromosome 17 (FTDP-17) is an inherited, progressive dementia with onset usually occurring between ages 40 and 60. Genetic testing for FTDP-17 is available from a few laboratories. Once an individual has been identified with the mutation, testing of siblings who may be age 65 or older may be appropriate for predispositional assessment of genetic risk status.

Pharmacogenetic tests are expected to become particularly relevant to older Medicare beneficiaries once the prescription drug benefit is implemented in 2006. Also, as we learn more about the genetic basis for common, chronic diseases, more predispositional genetic tests are expected to become available that are relevant to the elderly population.

Finally, there are many predispositional genetic tests that exist that could benefit younger beneficiaries, who make up 15 percent of the Medicare population.

CMS has a long-standing policy that states that "tests that are not reasonable and necessary for the diagnosis or treatment of an illness or injury are not covered"[38] and more specifically, that "tests for screening purposes that are performed in the absence of signs, symptoms, complaints, or personal history of disease or injury are not covered except as *explicitly authorized by statute*."[39] This policy is based largely on an interpretation of statutory language that allows for coverage of expenses that are "reasonable and necessary for the diagnosis of illness or injury or to improve the functioning of a malformed body member"[40] and that excludes coverage of expenses "for routine physical checkups, eyeglasses, or eye examination, …hearing aids or examination."[41] The preventive screening tests that Medicare does cover have been legislatively authorized by an act of Congress on a service-by-service basis.

Application of Screening Exclusion to Genetic Tests. Since CMS considers predictive and predispositional genetic tests to be screening tests and since coverage of such tests has not been explicitly authorized by Congress, they are not covered by Medicare. Diagnostic genetic tests, on the other hand, can be covered under Medicare because they are performed in the presence of signs and symptoms of disease. Pharmacogenetic tests that are performed in the presence of signs, symptoms, or a personal history of disease or adverse drug

[38] For example, see 66 FR 58813.
[39] 42 USC 1395y(a).
[40] 42 U.S.C. §1395y.

reactions could be covered under Medicare. However, it is not clear whether pharmacogenomic testing using microarrays (e.g., Roche's CYP450 AmpliChip) to test several different drug-metabolizing genes for a large variety of single nucleotide polymorphisms (SNPs) at one time would be covered since these genes and SNPs may not all be informative with respect to the treatment of the disease at hand. Pharmacogenomic testing used as a screening tool (in the absence of signs, symptoms, personal history, or complaints of disease) to help guide drug therapy decisions probably would not be covered under Medicare.

Application of Screening Exclusion to Genetic Counseling Services. The screening exclusion also limits Medicare coverage for genetic counseling services. Much of genetic counseling involves discussions with patients who have a strong family history of disease; however, family history of disease does not meet Medicare's reasonable and necessary criterion. Therefore, Medicare does not cover genetic counseling accompanying a predictive or predisposition genetic test in the absence of signs, symptoms, or personal history of disease.

Increasing Receptiveness to Coverage of Preventive Services. Recent discussions in both Congress and throughout HHS suggest that policymakers may be growing more receptive to the idea that Medicare should broaden its coverage of preventive services. In the past two decades, Congress has authorized Medicare coverage for several screening tests and services (e.g., mammography). Authorization for coverage of three new preventive services through the Medicare Prescription Drug, Modernization, and Improvement Act of 2003 (MMA) reflects an increasing focus on prevention. The MMA also authorized several demonstration projects in disease management. Given the potential role for preventive and/or diagnostic genetic tests in disease management, there may be an opportunity to include them in these ongoing demonstration projects.

Recommendation 4

Predictive and predispositional genetic tests can be clinically beneficial when there are no current signs, symptoms, or personal history of illness. As such, predictive and predispositional genetic tests and their accompanying services that meet evidence standards should be covered under Medicare.

The Secretary should urge Congress to add a benefit category for preventive services that would enable CMS to use its national coverage decisionmaking process, which includes an assessment of existing evidence, to determine whether a test or service is reasonable and necessary for the prevention or early detection of an illness or disability in asymptomatic individuals and, thus, ought to be covered. A statutory change would allow CMS to consider covering many more genetic tests and services used for preventive purposes.

More immediately, the Secretary should direct CMS to clarify that, in certain cases as scientific evidence warrants, a "personal history" of disease can include having a family history of a disease. This change would make it possible for a beneficiary with a family history of a disease to meet the "reasonable and necessary" standard for Medicare coverage. CMS will need to develop criteria that define when a family history should be considered a personal history of disease.

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[41] Ibid.

Medicaid Coverage

Medicaid Coverage of Genetic Tests and Services. The Federal Government mandates that certain benefits be provided to Medicaid recipients, and the States have discretion to cover additional benefits. With the exception of newborn screening and followup, which is State mandated, coverage of genetic tests and services are an optional Medicaid benefit. Limited data have been gathered on the extent of State Medicaid coverage of genetic tests and services, and States' Medicaid statutes are often broadly defined, making it difficult to determine with any certainty the extent to which genetic tests and services are covered under Medicaid.

State Variation. Because States are responsible for making coverage decisions for genetic tests and services, significant heterogeneity in Medicaid coverage across States can result. Such heterogeneity may constitute a significant source of disparate access to genetic tests and services, especially since Medicaid beneficiaries may not have other health insurance coverage or be able to pay for care out-of-pocket.

State revenues and the political climate are two factors that can affect a State's decisions to cover genetic tests and services. The size of a State's revenue stream and tax base can affect its ability to cover new medical technologies, including genetic tests, under Medicaid. States with higher tax rates and more generous welfare benefits may be more likely to cover new genetic tests and services under Medicaid. Coverage decisions in some States also may be affected by the fact that genetic tests are used for reproductive decisionmaking or family planning, which are viewed by some as tests that can lead to pregnancy termination. As of May 2004, 23 states use their own funds to cover abortion services under Medicaid (use of Federal funds for such services is prohibited).[42] If coverage for genetic tests performed for reproductive decisionmaking purposes follows the same pattern, access to such tests within the Medicaid population would be similarly uneven.

State Budgets. Unlike the Federal Government, which can legally operate with a deficit, all 50 States, by State law or custom, balance their budgets at the end of their fiscal year. States cannot create money in the way the Federal Government can, and thus, their tax revenues primarily determine spending levels. This key distinction in fiscal management translates into major differences in how Medicare and Medicaid operate. Economic difficulties since 2001 have affected most States, and their budgets have been extremely constrained as a result. As a means-tested welfare program, Medicaid represents possibly the second largest source of pressure on State budgets. Across States, Medicaid expenditures account for approximately 22 percent of all State spending.[43] In times of fiscal difficulty, states often reduce Medicaid expenditures by limiting coverage or payment rates to balance their budgets.

In contrast to Medicare, whose NCDs guarantee coverage for a service from that point forward (unless it is subsequently modified), Medicaid benefits that go beyond the Federal requirements can be scaled back or dropped at any time by States, and eligibility requirements can be made more stringent. The balanced budget mandate can create instability in the level of coverage States can provide for health services and tests. Even if a State decides to add a new genetic test or service to its benefit package one year, it could be dropped the next. States' fiscal policy thus makes it extremely difficult for the Medicaid population to secure access to new tests and services over the long term.

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[42] The Alan Guttmacher Institute, State Funding of Abortion under Medicaid, as of May 3, 2004. http://www.guttmacher.org/pubs/spib_SFAM.pdf [November 18, 2005].

[43] National Governor's Association and National Association of State Budget Officers. The Fiscal Survey of States. June 2005. http://www.nasbo.org/Publications/fiscalsurvey/fsspring2005.pdf [November 19, 2005].

Recommendation 5

The Secretary should ensure that States receive information about the existing evidence base and other supporting information about genetic tests and services, such as guiding principles that serve as the basis for coverage decisionmaking (see Recommendation 1). This information should be used by States to inform their Medicaid coverage decisions.

Through the provision of grant funding, HHS should continue to encourage States to cover, adopt, and provide genetic tests and services with a sound evidence base.

Billing and Reimbursement

Billing is the process by which a health provider or plan member submits a claim to a health insurance plan requesting reimbursement for the provision of a particular medical service. Once the claim is received, the health insurance plan reviews it to determine whether the medical service is covered under the member's health insurance plan and whether the circumstances under which the service was provided meet the criteria for coverage.[44] If all conditions are met, the claim is paid. In some instances, the health insurance plan may require the provider to supply additional documentation that justifies the need for the medical service. If a formal coverage policy or a case-based precedent for payment does not exist, a more detailed review may be necessary.

Reimbursement is the payment given to a provider or facility for medical services rendered to health insurance plan members. Reimbursement rates are frequently negotiated as one of the contract terms between health insurance plans and network providers. These negotiated fees are usually based on the usual, customary, and reasonable charges for that service and geographic area but are generally less than the amount billed to uninsured patients by providers. Providers usually are willing to accept discounted payment in exchange for a certain volume of patients that the health insurance plan can bring to the provider's practice. Because rates are negotiated on a contractual basis, reimbursement amounts for the same service can vary from provider to provider. Payment rates also vary by geographic area and the setting in which the service is provided.

Coding Systems

With millions of health insurance claims processed daily, efficient systems must be in place to facilitate these transactions. The Current Procedural Terminology (CPT) coding system, the Health care Common Procedural Coding System (HCPCS), and the International Classification of Diseases (ICD) have been developed to expedite this process. These coding systems are designed to convey information about the nature of services provided, the technologies used, and the patient's underlying illness. They help health insurance plans assess whether the care provided is covered by the patient's health plan, whether the circumstances warrant coverage, and, ultimately, whether the claim ought to be paid. Payment for medical services is based on the code(s) associated with a particular service and the dollar amount(s) assigned to the code(s).

If existing coding systems are not sufficiently descriptive of the service being provided and the reason it is being provided, it can be difficult for health insurance plans to process the claim appropriately and efficiently.

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[44] A health insurance plan may have other conditions that must be met for payment to be issued, such as meeting timeframes for submitting claims.

If claims for genetic tests and services are repeatedly denied because of inadequate codes, providers and laboratories may become less willing to offer these tests and services or to accept third-party reimbursement. In such situations, costs may be transferred to patients who are unable to pay for the test, and services may become difficult to access.

Current Procedural Terminology. Created in 1966 by AMA, the CPT coding system is a list of standard descriptive terms and identifying codes for reporting medical services and procedures to public and private health programs. CPT codes consist of a five-digit number that is associated with a brief description of the procedure.

CPT Editorial Panel. CPT codes are updated annually by the AMA CPT Editorial Panel. The CPT Editorial Panel is responsible for revising, modifying, and updating CPT codes as new and emerging tests are developed and replace outmoded procedures. The Panel will consider adding a new code or changing a code if it (1) is for a distinct clinical service performed throughout the United States, (2) is provided or supervised by physicians or other type of health professional, (3) and does not duplicate or fragment existing codes; and (4) if the clinical efficacy of the service is well established and documented in U.S. peer-reviewed literature. Panel decisions include (1) approval of a new code, (2) revision of existing nomenclature, (3) tabling of a proposal until further information is obtained, or (4) rejection of a request.[45]

Pathology Coding Caucus

The Pathology Coding Caucus was formed to increase participation by non-physician stakeholders in the development of CPT laboratory and pathology codes and review of code revision proposals. The group meets periodically to review proposed new codes, suggest revisions to existing codes, and develop consensus recommendations. Its membership consists of representatives from the American Medical Association (AMA), Advanced Medical Technology Association, American Association of Clinical Chemistry, American Clinical Laboratory Association, American Society for Clinical Pathology, American Society of Cytopathology, College of American Pathologists, National Association of Medical Examiners, U.S. & Canadian Academy of Pathology, Clinical Laboratory Management Association, American Society for Microbiology, and American Association of Bioanalysts.

CPT Codes for Genetic Tests. Genetic tests are billed using pathology and laboratory codes (see page 39 for listing of genetic test CPT codes). The CPT codes used for billing genetic tests are not specific to the condition being tested; rather, the code identifies the procedure performed (e.g., reverse transcription). Usually, several codes are used when billing for genetic testing that reflect the multiple steps involved in genetic testing.

Because CPT codes for genetic tests are procedure based, a new genetic test performed using existing procedures does not result in the development of a new CPT code. Thus, the payment amount for a new genetic test is based on the reimbursement amounts associated with existing CPT codes.

CPT Code Modifiers. The five-digit codes available for billing genetic tests and services had been criticized for not being specific enough to allow health insurance plans to make informed claim determinations, resulting in denials or requests for additional information. In response to this criticism, the Genetic Test

[45] American Medical Association. CPT Process: How a code becomes a code. http://www.ama-assn.org/ama/pub/category/3882.html [November 18, 2005].

Coding Workgroup,[46] a consortium of genetics and laboratory organizations, submitted a proposal to the CPT Editorial Panel requesting the addition of two-digit modifiers to supplement the existing five-digit CPT laboratory codes used for genetic testing. AMA adopted these modifiers and included them in Appendix I of CPT 2005, its annual listing of current CPT codes. The first numeric digit indicates the disease category, and the second alpha digit denotes gene type (see pages 40-42 for list of genetic testing code modifiers). These modifiers convey important information to health insurance plans about the nature of the test being billed so that they may better adjudicate claims. They will not, however, change the reimbursement rates associated with these codes.

It is anticipated that these modifiers will be less prone to payment denial and will allow for better tracking of utilization of genetic tests according to gene type and disease category. It is premature, however, to say whether the modifier codes will have their desired effect.

Category III CPT Codes. To facilitate the collection of data for new and emerging technologies and services, a separate set of CPT codes is available for data collection. These Category III codes are used to help substantiate widespread usage or obtain premarket approval by the Food and Drug Administration but are reimbursed less frequently because the associated technologies often are considered experimental. These codes are particularly appropriate for novel genetic technologies and testing procedures, but they are not useful for new genetic tests that use existing technologies and laboratory procedures for which CPT codes already exist.

CPT Codes for Genetic Counseling Services. Genetic counseling services are billed using evaluation and management (E&M) CPT codes. Like the genetic testing CPT codes, E&M codes used for billing genetic counseling services are not specific to genetic services; rather, they are generic codes used by all specialty types for patient visits. The codes are grouped into four categories: (1) consultation codes for patients referred by another physician; (2) office visit codes for self-referred patients; (3) preventive medicine/risk reduction codes for services provided to promote health and prevent illness or injury; and (4) health behavior and assessment codes for services associated with acute or chronic illness, prevention of a physical illness or disability, and maintenance of health provided by nonphysicians (see page 42 for list of genetic counseling CPT codes).

Determinations about which E&M code to use are based on a number of components. The extent of history obtained, extent of the physical examination, and complexity of medical decisionmaking are the key factors considered when selecting the E&M code level. When the visit consists predominantly of counseling and/or coordination of care, the amount of time spent face-to-face with the patient becomes the key factor for determining the code level.[47] The highest level E&M code available (level 5) is for an 80-minute visit (many genetic counseling visits can last 2 hours or longer).

Prolonged service codes are available when patient contact is longer than usual; however, obtaining reimbursement for these codes can be challenging. Using multiple codes or using codes multiple times for the same visit to account for the additional time spent is not permitted.

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[46] The Genetic Test Coding Workgroup is distinct from, but its membership overlaps with, the Pathology Coding Caucus described in box on page 40.

[47] Current Procedural Terminology 2005 © American Medical Association.

Planned Revisions to E&M Codes. The CPT Editorial Panel is currently considering proposals for the creation of CPT codes for family history/risk assessment/pedigree analysis for inclusion in CPT 2007.

E&M Code Relative Values. The AMA, through its Relative Value Scale Update Committee, assigns each E&M code a relative value unit. The relative value unit assigned to each E&M code is based on the time, effort, and technical skill it takes to perform the service as well as practice expense. The relative value units are updated each year to account for changes in medical practice.

CMS uses this relative value to determine Medicare payment rates for E&M codes. Medicare payment rates for E&M codes are calculated by multiplying the relative value by a monetary amount that is determined by CMS. These payment rates are then adjusted for geographical differences in resource costs.

Healthcare Common Procedural Coding System. The Healthcare Common Procedural Coding System (HCPCS) is a two-level system used for billing Medicare and Medicaid. Level I codes refer to CPT codes. Level II codes are five-digit, alpha-numeric codes that are not included in CPT but that identify services, supplies, and equipment that Medicare or Medicaid may cover. Examples of Level II Healthcare Common Procedural Codes (HCPCs) that are particular to genetic tests and services include complete and single-gene sequence analysis for breast and ovarian cancer, cystic fibrosis, hemochromatosis, and hereditary nonpolyposis colorectal cancer. The CMS HCPCS Workgroup, composed of representatives of the major components of CMS and the State Medicaid agencies, is responsible for making changes to HCPCs or their descriptors. Although the availability of an HCPC makes it possible to submit a claim for tests or services that lack a CPT code, it does not guarantee payment for that service.

International Classification of Diseases. Now in its 10th edition, the International Classification of Diseases (ICD) is a product of the World Health Organization. CMS and the Centers for Disease Control and Prevention publish a clinical modification of the ICD called ICD-9-CM for classification of causes of morbidity and mortality. ICD-9-CM codes are reported in combination with CPT codes or HCPCs when billing health plans to clarify the reason for referral or diagnosis for which the service is being provided. This information is used to help determine whether the plan member's circumstances meet coverage criteria. Like CPT codes, ICD codes also have undergone considerable revision to reflect suspected genetic diagnoses. "V" codes are the category of ICD-9-CM codes primarily used when billing genetic services provided to asymptomatic individuals. Health plans have sometimes been reluctant to reimburse V codes, and when they do, the reimbursement rate is often low.[48]

[48] National Society of Genetic Counselors. Primer on Billing and Reimbursement for Genetic Counselors. 2003.

Genetic Test CPT Codes	
83890	Molecular isolation or extraction
83891	Isolation of highly purified nucleic acid
83892	Enzymatic digestion
83893	Dot/slot blot production
83894	Separation by gel electrophoresis
83896	Nucleic acid probe, each
83897	Nucleic acid transfer (e.g., Southern, Northern)
83898	Amplification, single primer pair, each primer pair
83901	Amplification, multiplex, each reaction
83902	Reverse transcription
83903	Mutation scanning, single segment, each
83904	Sequencing, single segment, each
83905	Allele-specific transcription, single segment, each
83912	Interpretation and report
88230	Tissue culture for neoplastic disorders: lymphocyte
88233	Tissue culture for neoplastic disorders: skin or other solid tumor biopsy
88235	Tissue culture for neoplastic disorders: amniotic fluid or chronic villus cells
88237	Tissue culture for neoplastic disorders: bone marrow, blood cells
88239	Tissue culture for neoplastic disorders: solid tumor
88240	Cryopreservation, freezing and storage of cells, each aliquot
88241	Thawing and expansion of frozen cells, each aliquot
88245	Chromosome analysis for breakage syndrome: baseline sister chromatid exchange, 20-25 cells
88248	Chromosome analysis for breakage syndrome: baseline breakage, score 50-100 cells, count 20 cells, 2 karyotypes
88249	Chromosome analysis for breakage syndrome: score 100 cells, clastogen stress
88261	Chromosome analysis: count 5 cells, 1 karyotype with banding
88262	Chromosome analysis: count 15-20 cells, 2 karyotypes with banding
88263	Chromosome analysis: count 45 cells for mosaicism, 2 karyotypes with banding
88264	Chromosome analysis: analyze 20-25 cells
88267	Chromosome analysis: amniotic fluid or chorionic villus, count 15 cells, 1 karyotype with banding
88269	Chromosome analysis: in situ for amniotic fluid cells, count cells from 6-12 colonies, 1 karyotype with banding
88271	Molecular cytogenetics: DNA probe, each
88272	Molecular cytogenetics: chromosomal in situ hybridization, analyze 3-5 cells
88273	Molecular cytogenetics: chromosomal in situ hybridization, analyze 10-30 cells
88274	Molecular cytogenetics: interphase in situ hybridization, analyze 25-99 cells
88275	Molecular cytogenetics: interphase in situ hybridization, analyze 100-300 cells
88280	Chromosome analysis: additional karyotypes, each study
88283	Chromosome analysis: additional specialized banding technique
88285	Chromosome analysis: additional cells counted, each study
88289	Chromosome analysis: additional high-resolution study
88291	Interpretation and report
88299	Unlisted cytogenetic study

CPT Genetic Testing Code Modifiers	
Neoplasia (solid tumor)	
-0A	BRCA1 (hereditary breast/ovarian cancer)
-0B	BRCA2 (hereditary breast cancer)
-0C	neurofibromin (neurofibromatosis, type 1)
-0D	merlin (neurofibromatosis, type 2)
-0E	c-RET (multiple endocrine neoplasia, types 2A/B, familial medullary thyroid carcinoma)
-0F	VHL (von Hippel-Lindau disease)
-0G	SDHD (hereditary paraganglioma)
-0H	SDHB (hereditary paraganglioma)
-0I	her-2/neu
-0J	MLH1 (hereditary nonpolyposis colorectal cancer)
-0K	MLH2 (hereditary nonpolyposis colorectal cancer)
-0L	APC (hereditary polyposis coli)
-0M	Rb (retinoblastoma)
-1Z	solid tumor, not otherwise specified
Neoplasia (lymphoid/hematopoetic)	
-2A	AML1 – also ETO (acute myeloid leukemia)
-2B	BCR – also ABL (chronic myeloid, acute lymphoid leukemia)
-2C	CGF 1
-2D	CBFBeta (leukemia)
-2E	MML (leukemia)
-2F	PML/RARalpha (promyelolocytic leukemia)
-2G	TEL (leukemia)
-2H	bcl2 (lymphoma)
-2I	bcl1 (lymphoma)
-2J	c-myc (lymphoma)
-2K	IgH (lymphoma/leukemia)
-2Z	lymphoid/hematopoetic neoplasia, not otherwise specified
Non-neoplastic hematology/coagulation	
-3A	Factor V (leiden, others)(hypercoagulable state)
-3B	FACC (Fanconi anemia)
-3C	FACD (Fanconi anemia)
-3D	beta globin (thalassemia)
-3E	alpha globin (thalassemia)
-3F	MTHFR (elevated homocysteine)
-3G	prothrombin (Factor II, 20210A)(hypercoagulable state)
-3H	Factor VII (hemophilia A/VWF)
-3I	Factor XI (hemophilia B)
-3J	beta globin
-3Z	non-neoplastic hematology/coagulation, not otherwise specified

CPT Genetic Testing Code Modifiers (continued)	
Histocompatability/blood typing	
-4A	HLA-A
-4B	HLA-B
-4C	HLA-C
-4D	HLA-D
-4E	HLA-DR
-4F	HLA-DQ
-4G	HLA-DP
-4H	Kell
-4Z	histocompatability/blood typing, not otherwise specified
Neurologic, non-neoplastic	
-5A	aspartoacylase A (Canavan disease)
-5B	FMR-1 (Fragile X, FRAXA syndrome)
-5C	frataxin (Friedreich ataxia)
-5D	huntingtin (Huntington disease)
-5E	GABRA (Prader-Willi/Angelman syndrome)
-5F	connexin-26 (GJB2)(hereditary deafness)
-5G	connexin-32 (X-linked Charcot-Marie-Tooth disease)
-5H	SNRPN (Prader-Willi/Angelman syndrome)
-5I	ataxin-1 (spinocerebellar ataxia, type 1)
-5J	ataxin-2 (spinocerebellar ataxia, type 2)
-5K	ataxin-3 (spinocerebellar ataxia, type 3, Machado-Joseph disease)
-5L	CACNA1A (spinocerebellar ataxia, type 6)
-5M	ataxin-7 (spinocerebellar ataxia, type 7)
-5N	PMP-22 (Charcot-Marie-Tooth disease, type 1A)
-5O	MECP2 (Rett syndrome)
-5Z	neurologic, non-neoplastic, not otherwise specified
Muscular, non-neoplastic	
-6A	dystrophin (Duchenne/Becker muscular dystrophy)
-6B	DMPK (myotonic dystrophy, type 1)
-6C	ZNF-9 (myotonic dystrophy, type 2)
-6D	SMN (autosomal recessive spinal muscular atrophy)
-6Z	muscular, not otherwise specified
Metabolic, other	
-7A	apolipoprotein E (cardiovascular disease, Alzheimer's disease)
-7B	sphingomyelin phosphodiesterase (Niemann-Pick disease)
-7C	acid beta glucosidase (Gaucher disease)
-7D	HFE (hemochromatosis)
-7E	hexosaminidase A (Tay-Sachs disease)
-7Z	metabolic, not otherwise specified
Metabolic, transport	
-8A	CFTR (cystic fibrosis)
-8Z	metabolic, transport, not otherwise specified

CPT Genetic Testing Code Modifiers (continued)	
Metabolic-pharmacogenetic	
-9A	TPMT (thiopurine methyltransferase (patients on antimetabolite therapy)
-9L	metabolic-pharmacogenetics, not otherwise specified
Dysmorphology	
-9M	FGFR1 (Pfeiffer and Kallman syndromes)
-9N	FGFR2 (Crouzon, Jackson-Weiss, Apert, Saethre-Chotzen syndromes)
-9O	FGFR3 (achondroplasia, hypochondroplasia, thanatophoric dysplasia, types I and II, Crouzon syndrome with acathosis nigricans, Muenke syndromes)
-9P	TWIST (Saethre-Chotzen syndrome)
-9Q	CATCH-22 (22q11 deletion syndromes)
-9Z	dysmorphology, not otherwise specified)

Genetic Counseling CPT Codes	
96150-96155	Health and behavioral assessment
99078	Group education sessions
99201-99205	Office/outpatient service – new patient self-referred, 10-60 minutes
99211-99215	Office/outpatient service – established patient
99241-99245	Office/outpatient consultations – new or established patients, 15-80 minutes
99251-99255	Initial inpatient consultations
99261-99263	Follow-up inpatient consultations
99271-99275	Confirmatory consultations – new or established patients
99360	Physician standby services requiring prolonged physician attendance without direct (face-to-face) patient contact
99361-99362	Medical conference to coordinate activities of patient care, 30-60 minutes
99381-99387	Preventive medicine – new patient
99391-99397	Preventive medicine – established patient
99401-99404	Individual counseling of risk factor reduction for healthy individual, 15-60 minutes
99411-99412	Group counseling of risk factor reduction for healthy individuals, 30-60 minutes
99420	Administration and interpretation of health risk assessment
99499	"Incident to" physician
Modifier-21	Prolonged service (added to five-digit code when face-to-face or floor/unit service provided is prolonged or otherwise greater than that usually required for the highest level of E&M service within a given category)
Facility Codes:	
99121-99125	Established patients
99201-99205	New patients
99242-99245	Consultations
99272-99275	Conference consultations

Medicare Clinical Laboratory Fee Schedule

Rather than negotiating payment rates with each individual provider, some health insurance plans opt to set a fee schedule that lists the maximum amount that it will pay for particular services regardless of who is providing the service and where it is being provided. These rates may be adjusted for geographic location or other factors deemed relevant by the health insurance plan. Medicare has a fee schedule for clinical laboratory services that has been used by health insurance plans to determine their own payment levels and often as a basis for negotiating discounted rates. As a result, any deficiencies in Medicare's fee schedule can extend to private insurance.

Established in the early 1980s, the Medicare Clinical Laboratory Fee Schedule is a list of the amounts that Medicare will reimburse for clinical laboratory procedures, including genetic tests. For each procedure, the amount reimbursed is the lowest among the submitted charge, national limit amount, or local fee schedule amount. Local Medicare administrative contractors may set the local fee schedule amount at the national limit or at a lower rate.

Payment rates for a new laboratory test assigned a new code are handled through two methods: gap-filling and cross-walking. Gap-filling is a decentralized process that involves asking local Medicare administrative contractors to determine a local payment amount for the new code. The payment amount is based on charges for the test resources required to perform the test, clinical study findings, and information from local clinicians, manufacturers, and other interested parties. These local payment rates then are used to set the national limit amount for subsequent years. Cross-walking refers to the process by which a new laboratory code is assigned the same fee as an existing code for a similar lab test or procedure. Fees for new laboratory codes have been established primarily through the cross-walking process.[49]

"Fee Freeze." The fee schedule's authorizing legislation calls for annual updates; however, the recent Medicare Prescription Drug, Improvement, and Modernization Act of 2003 (MMA) imposed a fee freeze on payment rates for clinical laboratory tests, locking rates at the 2003 level until 2009, despite inflation and increasing labor and technology costs to laboratories to provide testing.[50] This new fee freeze follows a previous 5-year moratorium that ended in 2002. As a result, Medicare laboratory fees have not kept pace with inflation as well as changes in general laboratory costs and technological advances that have improved efficiency and/or quality.[51]

Revising the Medicare Clinical Laboratory Fee Schedule. Many providers that bill Medicare contend that current fees do not reflect a genetic test's true cost. Although the laboratory fee schedule may be revisited and comprehensively revised after 2009, currently there is no mechanism available for addressing concerns about underreimbursement through comprehensive revision of the entire fee schedule. Even if it were possible, a complete revision of the laboratory fee schedule might not resolve these concerns. Also, older, higher volume tests, whose payment rates reflect the cost when the test was first established and

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[49] Raab GG and Logue LJ. Medicare coverage of new clinical diagnostic laboratory tests: The need for coding and payment reforms. *Clin Leadership Management Rev* 2001. 15(6):376-87.
[50] Public Law 108-173. Medicare Prescription Drug, Improvement, and Modernization Act of 2003.
[51] Institute of Medicine. Medicare Laboratory Payment Policy: Now and in the Future. 2000. http://www.nap.edu/books/0309072662/html/ [November 18, 2005].

the technology to perform it was perhaps more costly, can subsidize newer, more costly tests that may be underreimbursed. Any comprehensive revision of the fee schedule could result in an increase in fees for newer tests and a decrease in fees for older tests that can now be performed at a lower cost.

2005-2008 Medicare Clinical Laboratory Fee Schedule Amounts for Genetic Testing Codes

HCPCS Code	Description	State Median Limit	Range	National
83890	Molecular isolation or extraction	$5.60	$3.26-5.60	$7.57
83891	Isolation of highly purified nucleic acid	$5.60	$3.26-5.60	$7.57
83892	Enzymatic digestion	$5.60	$3.26-5.60	$7.57
83893	Dot/slot blot production	$5.60	$3.26-5.60	$7.57
83894	Separation by gel electrophoresis	$5.60	$3.26-5.60	$7.57
83896	Nucleic acid probe, each	$5.60	$3.26-5.60	$7.57
83897	Nucleic acid transfer (e.g., Southern, Northern)	$5.60	$3.26-5.60	$7.57
83898	Amplification, single primer pair, each primer pair	$23.42	$5.37-23.42	$31.65
83901	Amplification, multiplex, each reaction	$23.42	$5.37-23.42	$31.65
83902	Reverse transcription	$19.83	$5.47-19.82	$26.80
83903	Mutation scanning, single segment, each	$23.42	$5.37-23.42	$31.65
83904	Sequencing, single segment, each	$23.42	$5.37-23.42	$31.65
83905	Allele-specific transcription, single segment, each	$23.42	$5.37-23.42	$31.65
83912	Interpretation and report	$5.60	$3.26-5.60	$7.57
88230	Tissue culture, lymphocyte	$162.77	$28.46-162.77	$162.77
88233	Tissue culture, skin/biopsy	$196.63	$33.97-196.63	$196.63
88235	Tissue culture, placenta	$205.74	$33.97-205.74	$205.74
88237	Tissue culture, bone marrow	$176.47	$28.46-176.47	$176.47
88239	Tissue culture, tumor	$206.12	$28.71-206.12	$206.12
88240	Cell cryopreserve/storage	$14.11	$5.93-14.11	$14.11
88241	Frozen cell preparation	$14.11	$5.93-14.11	$14.11
88245	Chromosome analysis: 20-25 cells	$207.98	$89.29-207.98	$207.98
88248	Chromosome analysis: 50-100 cells	$241.96	$124.89-241.96	$241.96
88249	Chromosome analysis: 100 cells	$241.96	$124.89-241.96	$241.96
88261	Chromosome analysis: 5 cells	$246.93	$235.33-246.93	$246.93
88262	Chromosome analysis: 15-20 cells	$174.14	$174.14-174.14	$174.14
88263	Chromosome analysis: 45 cells	$209.97	$89.29-209.97	$209.97
88264	Chromosome analysis: 20-25 cells	$174.14	$174.14-174.14	$174.14
88267	Chromosome analysis: placenta	$251.17	$211.13-251.17	$251.17
88269	Chromosome analysis: amniotic	$232.38	$169.34-232.38	$232.38
88271	Cytogenetics: DNA probe	$29.93	$16.84-29.93	$29.93
88272	Cytogenetics: 3-5 cells	$37.41	$25.28-37.41	$37.41
88273	Cytogenetics: 10-30 cells	$44.89	$30.33-44.89	$44.89
88274	Cytogenetics: 25-99 cells	$48.63	$30.33-48.63	$48.63
88275	Cytogenetics: 100-300 cells	$56.11	$30.33-56.11	$56.11
88280	Chromosome karyotype study	$35.07	$35.07-35.07	$35.07
88283	Chromosome banding study	$35.07	$35.07-35.07	$35.07
88285	Additional chromosome count	$26.54	$7.54-26.54	$26.54
88289	Additional chromosome study	$48.11	$4.45-48.11	$48.11

Source: CMS. 2005 Clinical Laboratory Fee Schedule. http://new.cms.hhs.gov/ClinicalLabFeeSched/02_clinlab.asp#TopOfPage.

Inherent Reasonableness. HHS has the authority (known as "inherent reasonableness" authority) to revise payment levels for items or services when they are so grossly excessive or deficient that they threaten to reduce beneficiary access to care or represent a misappropriation of taxpayer dollars. A review for inherent reasonableness might be initiated when:

- A limited number of suppliers offer the service
- Medicare is the sole or primary source of payment for a service
- The payment amount does not reflect changing technology or changes in acquisition, production, or supplier costs
- The payment amount is grossly higher or lower than production costs
- There have been increases in payment amounts for a service that cannot be explained by inflation or technology
- The local payment amount is grossly higher or lower than the payment amount in other localities
- The local payment amount is grossly higher or lower than the local payment provided by other purchasers in the same locality[52]

Inherent reasonableness could provide a way to address instances of extreme underreimbursement and payment variation among localities for genetic test codes. However, the review process requires a significant amount of data collection and analysis and may result in a lower rather than a higher payment rate. Nonetheless, this mechanism might be used to temporarily redress extreme discrepancies between cost and payment for the CPT codes used for genetic tests until the laboratory fee schedule freeze is lifted.

Recommendation 6

When the congressional freeze on laboratory payment rates end in 2009, the Secretary should be prepared to revise payment rates to reflect the true cost of a genetic test. In the meantime, the Secretary should direct CMS to invoke its inherent reasonableness authority to address variations in payment rates for the genetic test CPT codes.

Clinical Diagnostic Laboratory Test Regulations. In an effort to increase the transparency of its rate setting process for new clinical diagnostic laboratory tests and to comply with Section 942(b) of the Medicare Drug Prescription, Improvement, and Modernization Act of 2003, Medicare will be establishing regulations that outline "procedures for determining the basis for, and amount of, payment under this subsection for any clinical diagnostic laboratory test with respect to which a new or substantially revised HCPC is assigned on or after January 1, 2005."[53] These regulations will not affect already existing HCPCs, including those currently used to bill for genetic tests.

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[52] Testimony of Thomas A. Scully, Administrator, Centers for Medicare & Medicaid Services. Medicare Payment for Medical Supplies. Senate Appropriations Labor, Health and Human Services, and Education Subcommittee. June 12, 2002. http://new.cms.hhs.gov/apps/media/press/testimony.asp?Counter=635 [January 4, 2006].

[53] Public Law 108-173.

Billing and Reimbursement for Genetic Tests

Payment Rates for Genetic Tests. Like coverage policies, payment rates are considered proprietary, making it difficult to empirically assess the adequacy of reimbursement for genetic tests and services. However, Medicare's Clinical Laboratory Fee Schedule, which is available to the public, is used by health plans throughout the United States as a baseline for the development and negotiation of their own fee schedules, making it a logical resource for comparing payment rates with actual costs for genetic tests in both the public and private sector.

In 2002 Medicare reported spending $13 million on 270,000 claims for genetic tests (approximately $48 per test).[54] Testimony provided to SACGHS in March 2004 showed that Medicare payment rates were significantly lower than the actual costs incurred by laboratories to provide genetic testing.[55] For instance, genetic testing for Fragile X syndrome, the most common inherited form of mental retardation, cost an academic laboratory in Virginia $266 to perform, but reimbursement is only $62. Similarly, genetic testing for Factor V Leiden, the most common hereditary blood coagulation disorder in the United States, costs $72 to perform, but reimbursement is only $68. Furthermore, major health plans, including Medicare and Medicaid, reimbursed genetic test claims only 60 to 90 percent of the time at this laboratory. The remaining 10 to 40 percent of test claims were not reimbursed at all or were reimbursed only partially. Unpaid costs were transferred to patients, who had to pay out-of-pocket, or were absorbed by the ordering physician or laboratory as a financial loss. Such financial losses provide a disincentive for laboratories to develop and offer genetic tests, which threaten to limit their availability to patients and overall integration into the health care system.

To minimize financial loss, some laboratories require confirmation of coverage from the patient's health insurance plan or full payment before they will perform the genetic test.[56] Obtaining prior authorization or advance payment can require significant time and effort by the patient and/or provider. In instances where the patient's health insurance plan will not pay the total charge, the outstanding charge is billed to the patient. This amount can be prohibitively expensive depending on the cost of the test and the insurer's reimbursement rate.

Balance billing of Medicaid beneficiaries is illegal but allowed by Medicare and some health insurance plans if the patient signs a waiver. Because biological samples are usually collected at a site different from the reference laboratory, obtaining such waivers can be difficult. Some providers/facilities will not send samples to laboratories that have such requirements for genetic testing, which can limit patient access to some genetic tests.

Some larger laboratories have established processes for securing adequate payment for genetic tests. LabCorp, for instance, informs health insurance plans about a new genetic test before or at the time it is

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[54] Sean Tunis, Medicare Coverage and Genetic Testing. Presentation to SACGHS on March 1, 2004. http://www4.od.nih.gov/oba/SACGHS/meetings/March2004/Tunis.ppt [November 18, 2005].

[55] Andrea Ferreira-Gonzalez, Providers' Perspective on Reimbursement of Genetic Tests and Services: A Laboratorian's Perspective. Presentation to SACGHS on March 1, 2004. http://www4.od.nih.gov/oba/SACGHS/meetings/March2004/Gonzalez.ppt [November 18, 2005].

[56] Myriad, for instance, follows this practice for its BRCA1/2 test (Myriad Reimbursement Assistance Program. http://www.myriadtests.com/mrap.htm [November 18, 2005]).

launched and provides them with comprehensive peer-reviewed documentation of clinical utility, evidence of improved health outcomes, cost assessments, professional society endorsements or comments, Federal agency opinions or recommendations, CMS coverage decisions, and public/political mandates to assist in the plans' technology review processes. It discusses with the health insurance plans what will satisfy the medical necessity requirement for test reimbursement, negotiates payment amounts, and resolves any reimbursement issues. It also verifies that (an) existing CPT code(s) can be used for billing or, if necessary, applies for (a) new code(s). Additionally, LabCorp develops educational materials for physicians and patients to explain the new test as well as advanced beneficiary notices to prepare patients for potential out-of-pocket costs they might incur if the test cost is not reimbursed by their health insurance plan. Once coverage and payment are established, the company will monitor payment and denial trends to correct any deficiencies in payment and resolve issues related to the medical necessity of testing.[57] Although such efforts improve the likelihood of adequate payment, problems such as denial of or partial payment persist.

Royalty Fees. Royalty fees can represent a sizable portion of genetic testing costs. These fees must be paid by the laboratory to the gene patent holder with whom they have a license to offer the test. The terms of these licenses often require a one-time, upfront flat fee that grants the laboratory permission to perform the tests as well as a fee for each test the laboratory performs. For example, Bio-Rad requires a $20 per test fee for its hereditary hemochromatosis genetic test in addition to an upfront payment inversely proportional to the laboratory's testing volume.[58] Additional examples include $12.50 per test for Canavan disease, $5 per test for Gaucher disease, and $2 per test for cystic fibrosis when the annual testing volume exceeds 750.[59] Because there is no CPT code that allows laboratories to seek separate reimbursement for royalty payments, these fees must be taken from the payments received from health plans and/or patients or absorbed by the laboratory conducting the test.

Because most licenses contain a confidentiality clause that prevents disclosure of the terms of the agreement, empirical data on the extent to which royalty fees comprise the cost of the test and their impact on access are difficult to obtain. At the academic laboratory discussed above, 9 to 15 percent of payment goes toward royalty fees. Another example cited in the literature indicates that royalty payments make up 20 percent of the cost (approximately $100) for a panel of tests for Tay-Sachs disease, cystic fibrosis, Gaucher disease, Niemann-Pick disease, and Canavan disease.[60] In a survey conducted by Cho et al. in 2001, laboratory directors reported that the costs of genetic testing to the laboratories and patients had increased because of patenting and licensing practices.[61] Combined with low reimbursement rates, these royalty fees could have a chilling effect on the availability of genetic testing services, especially for panel assays or multiplex genetic tests that screen many genes covered by multiple patents.[62]

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[57] Billings PR and Karnes T. Reimbursement for genetic testing: Issues noted by Laboratory Corporation of America Holdings, Inc. Submitted as public comment to SACGHS in September 2004.
[58] Merz JF et al. Diagnostic testing fails the test. *Nature* 2002. 415:577-9.
[59] Ibid.
[60] Ibid.
[61] Cho MK et al. Effects of patents and licenses on the provision of clinical genetic testing services. *J Mol Diagn* 2003. 5(1): 3-8.
[62] Prepared for AdvaMed by the Lewin Group. The Value of Diagnostics: Innovation, Adoption and Diffusion into Health Care. July 2005. http://www.advamed.org/publicdocs/thevalueofdiagnostics.pdf [November 18, 2005].

Medicaid Billing and Reimbursement. Medicaid payment rates are not permitted to exceed the national limit amount of the Medicare Clinical Laboratory Fee Schedule. In addition, many States require that samples be processed by in-State laboratories. If an in-State laboratory is not available, prior authorization can be obtained for the out-of-State laboratory to be reimbursed. Medicaid beneficiaries may have difficulty accessing certain genetic tests, especially those for rare diseases or those performed by only one or a few laboratories in the country because of gene patent or licensing restrictions, depending on the location of the laboratory relative to the State in which they reside. For example, one physician noted in her public comment that she has had difficulty in obtaining genetic testing for her Medicaid patients because some laboratories are less willing to accept the low Medicaid reimbursement rates of other States.[63] Because of their financial situations, Medicaid beneficiaries often are unable to pay out-of-pocket for the test and, therefore, may have to forgo recommended genetic testing.

Impact of Payment Rates for Genetic Tests. Health plans state that their reimbursement rates are based on billing patterns and provider charges. Manufacturers and providers, on the other hand, argue that many services and treatments are reimbursed at rates that are insufficient to cover the cost of the service and royalty fees or to recoup research and laboratory expenses.

From the perspective of the laboratory or manufacturer offering genetic testing services, inadequate payment rates can potentially threaten a laboratory's willingness to develop and offer genetic tests if they are provided at a financial loss, potentially limiting the availability of genetic tests to patients. These consequences of underreimbursement can be particularly problematic for individuals seeking testing for a rare genetic condition in which testing is available only from a single laboratory in the United States.

To guarantee reimbursement, laboratories that continue to offer genetic tests may choose to accept payment only from the patient rather than from the patient's health plan or require the patient to pay the difference between the laboratory's charge for the test and the amount the patient's health plan is willing to pay. If testing costs are transferred to the patient, access to genetic testing will be based on who can afford to pay out-of-pocket rather than who needs testing. If underreimbursement persists as genetics and genomics continue to play greater roles in the practice of medicine, the impact of low payment rates for genetic tests will intensify and will be experienced by a greater number of patients.

On the other hand, low reimbursement rates are a common complaint throughout the health care system. The problem is not unique to genetic tests. Furthermore, current reimbursement rates may act as an incentive to manufacturers and laboratories to develop more cost-efficient genetic technologies that will reduce the cost of testing.

Evidence-Based Payment Decisions. Market utilization data can help determine a fair and accurate reimbursement rate. Utilization data on new genetic tests can be difficult to obtain, however, because data cannot be captured unless the tests are reimbursed. Furthermore, new technologies, particularly when first implemented, often are more expensive than older technologies. Without sufficient reimbursement in place to guarantee utilization and long-term data collection, it can be difficult to demonstrate that a new genetic technology is actually clinically better than an existing one.

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[63] Public comments to SACGHS on draft Report on Coverage and Reimbursement of Genetic Tests and Services. June 2005.

Billing and Reimbursement for Genetic Counseling Services

Genetic counseling services often accompany predictive or predispositional genetic testing. They can be provided prior to testing to collect and interpret family, genetic, medical, and psychosocial information as well as to inform the patient of the various ethical, legal, and psychosocial issues raised by genetic testing. After a test is administered, genetic counseling services may be provided to discuss test results and the options of the patient based on those results.

A wide range of health professionals may provide genetic counseling services, including physicians, PhD medical geneticists, genetic counselors, physician assistants, nurses, clinical psychologists, family therapists, and clinical social workers. Certain providers of genetic counseling services will be more appropriate than others depending on the nature of the test and the condition for which the test is performed, the indications for testing, the complexity of the issues being discussed, and the education and qualifications of the provider. Providers less comfortable with offering genetic counseling services often refer their patients to genetics health providers who have specialized education and training or rely on their assistance for proper interpretation of test results.

Genetic counseling is distinct from other types of education and counseling in that it requires specific knowledge about genetics and inheritance. In addition to MD geneticists, several nonphysician providers are uniquely qualified to provide genetic counseling services because of their specialized training and certification. For example, genetic counselors are certified by the American Board of Genetic Counseling (ABGC), which requires graduation from an accredited genetic counseling graduate program and documentation of 50 cases to be eligible for initial certification and recertification every 10 years.[64] The Genetic Nursing Credentialing Commission (GNCC) offers an advanced practice nurse in genetics credential, which is available to registered nurses with at least a master's degree in nursing or equivalent and who have at least 3 years of experience as a genetic nurse with 50 percent of their practice involving genetic cases; at least 50 hours of patient contact with genetic content in the past 5 years; and 50 documented cases in the past 5 years.[65] GNCC also offers a genetics clinical nurse credential for nonmaster's nurses.[66] PhD medical geneticists who are certified by the American Board of Medical Genetics must demonstrate skills in interviewing and counseling "to elicit from the patient or family the information necessary to reach an appropriate conclusion, anticipate areas of difficulty and conflict, help families and individuals recognize and cope with their emotional and psychological needs, recognize situations requiring psychiatric referral, and transmit pertinent information in a comprehensible way to the individual or family."[67]

As the number of clinically relevant genetic tests rises, ensuring access to both genetic tests and counseling services will become increasingly important. Patients' access to genetic counseling services may be limited by a provider's inability to obtain a provider identifier number, lack of licensure, or inadequate payment

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[64] American Board of Genetic Counseling. 2005 Certification Examination Program. http://www.abgc.net/genetics/abgc/abgc-cert/2005/step-02.htm [November 18, 2005].
[65] Genetic Nursing Credentialing Commission. Advanced practice nurse in genetics requirements and information. http://www.geneticnurse.org/APNG.htm [November 18, 2005].
[66] Genetic Nursing Credentialing Commission. Genetic clinical nurse requirements and information. http://www.geneticnurse.org/APNG.htm [November 18, 2005].
[67] ABMG description of specialties in medical genetics. http://www.abmg.org/genetics/abmg/99-14.htm#phdmedgen [November 18, 2005].

and CPT codes. Generally, nonphysician providers have more difficulty securing reimbursement for genetic counseling services and, thus, are the focus of the following discussion.

Billing for Genetic Counseling Services. Payment rates for genetic counseling services can vary depending on how they are billed. Genetic counseling services can be billed either using generic CPT E&M codes or, to provided by a hospital employee, as part of the hospital facility fee. The lack of specific codes for genetic counseling services can be problematic due to the nature of these services as compared with regular provider visits. Specifically, genetic counseling sessions can last for 2 to 3 hours, not including the extensive time often spent preparing for a counseling session and following up with the patient.[68] However, the highest available CPT E&M code accounts for a significantly shorter timeframe. Prolonged service codes are available but are rarely reimbursed.

Billing Medicare. Under Federal regulations, not all genetic counseling providers are eligible to bill Medicare directly. Currently, physicians and some nonphysician practitioners—specifically, nurse practitioners, physician assistants, certified nurse specialists, certified nurse midwives, clinical psychologists, and clinical social workers—are statutorily eligible to bill Medicare directly.[69] Other nonphysician provider types not listed are not permitted to bill Medicare directly.

Auxiliary personnel provide care under a physician's direct supervision and bill Medicare for their services as "incident to" the supervising physician or hospital. Auxiliary personnel include, but are not limited to, nurses, nonphysician anesthetists, psychologists, technicians, therapists, genetic counselors, and other aides. Direct supervision of auxiliary personnel requires the physician to be present in the office suite and be immediately available to furnish assistance and direction (but the physician does not need to be present in the room) throughout the performance of the procedure.[70]

When billing Medicare for genetic counseling services provided by auxiliary personnel, the physician may utilize only the 99211 E&M code (used for billing for 5-minute visits with minimal problems present).[71]

Billing Private Health Insurance Plans. The ability of health providers to bill private health plans directly depends on the provider's scope of practice and employer, the policies of the health plan, and the State in which the service is provided. Physicians generally are able to bill directly for genetic services as long as they fall within their scope of practice. Nonphysician providers generally bill through a supervising physician unless the State allows their profession to practice independently and health plans permit them to bill directly for their services. Fifty-seven percent of genetic counselors responding to the 2004 Genetic Counselor Professional Status Survey conducted by the National Society of Genetic Counselors reported billing through their supervising physician, 9 percent reported billing in their and their supervising physician's name, and 2 percent reported billing in their name only.[72]

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[68] Preparation and followup time can involve constructing a family pedigree, reviewing medical records, conducting literature reviews, consulting practice guidelines, locating and contacting test laboratories, preparing or obtaining informational or educational materials to give to the patient, and finding information about support services.
[69] 42 CFR Ch. IV§410.20.
[70] Medicare Carrier Manual Section 2050.1(B).
[71] Personal communication with CMS official.
[72] National Society of Genetic Counselors, 2004 Professional Status Survey. http://www.nsgc.org/careers/2004_PSS_Final_pw.pdf [November 18, 2005].

Services of nonphysician providers employed by a hospital may be billed as part of the hospital's facility fee or as part of their unit's comprehensive fee, which is determined as part of the hospital's contract with the health plan. Eight percent of genetic counselors reported billing through the facility fee, and another 6 percent reported having their services included in the comprehensive fee.[73]

Billing Medicaid. State Medicaid programs' billing practices are similar to Medicare and private health insurance plans; however, some States have taken steps to facilitate direct billing by certain nonphysicians. For example, the State of Washington's Medicaid and Maternal Child Health Programs have begun credentialing genetic counselors as service providers so they can bill the State's Medicaid program directly for their services. In Ohio, board-certified genetics providers have been deemed by the state's Medicaid program as appropriate providers of medical genetic services and may bill Medicaid directly for their services.

Provider Identifiers. All physicians and specified nonphysician practitioners allowed to bill Medicare directly are required to have a Unique Provider Identifier Number (UPIN). Because auxiliary personnel are not permitted to bill Medicare directly for their services, they are not eligible for a UPIN.

In 1996 the Health Insurance Portability and Accountability Act (HIPAA) mandated the adoption of a uniform unique health provider identifier for all health providers, not just Medicare providers, for use in standard electronic transactions. In compliance with the HIPAA administrative simplification provisions and in an effort to remedy the limitations of the UPIN system, CMS is expected to implement the National Provider System (NPS) incrementally as a replacement to the UPIN system. Once NPS is implemented in 2006, all public and private health plans, health care clearinghouses, and health providers who are licensed, certified, or otherwise authorized to perform medical services or medical care, equipment, and/or supplies in the normal course of business must begin using the National Provider Identifier (NPI) (small health plans will have an additional year to comply). Auxiliary personnel (e.g., nurses, genetic counselors) who currently are not eligible for a UPIN but who are able to bill some health insurance plans directly will be eligible to receive an NPI. However, NPI eligibility would neither affect auxiliary personnel's current ineligibility to be directly reimbursed by Medicare nor guarantee payment for genetic counseling services.

State Licensure. The purpose of licensure is to ensure the high quality and safety of health services. In many States, only health providers licensed by the State may legally bill for health services, and many health insurance plans require health providers to be licensed to be credentialed as one of their network providers.

State licensure programs are available for most health professions that provide genetic counseling services, with the exception of genetic counselors. Currently, only three states—Illinois, California, and Utah—have passed legislation authorizing licensure of genetic counselors (only Utah has implemented the law), and 10 other States have introduced bills or are in the process of drafting bills that would establish licensure for genetic counselors.[74] Some of the reasons cited for not enacting licensure for genetic counselors include lack of

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[73] Ibid.

[74] Colorado, Florida, Massachusetts, New York, New Jersey, Oklahoma, Texas, Washington, and Wisconsin are currently considering or drafting bills that would enable licensure of genetic counselors.

evidence demonstrating harm to consumers in the absence of licensure, insufficient evidence demonstrating a need for licensure, concerns that other practitioners could be adversely affected or that patient access to genetic counseling providers would become restricted or reduced, and in some States with few genetic counselors, the high programmatic costs relative to the number of genetic counselors in the State. Although State licensure does not guarantee reimbursement,[75] it is expected to improve genetic counselors' ability to be recognized as qualified providers of genetic counseling services and, thus, to increase their prospects of being reimbursed for their services. Preliminary evidence from Utah has credited the establishment of licensure with increased recognition of the profession by payers in terms of being allowed to bill incident to a physician, fewer payment denials, and an increase in the State's genetic counseling workforce.[76]

Impact on Patient Access. The problems described above can adversely affect patients' access to genetic services. Several non-physician genetic counseling providers reported having difficulty securing and maintaining employment as a result of restrictions on their ability to bill for their services and difficulty generating sufficient revenue to support their salaries.[77] Some providers noted that they have had to reduce their patient caseload to devote more time to research that generates grant funding, which helps cover their salaries. These problems can lead to fewer qualified providers of genetic counseling services.

Several commenters expressed concern that these trends, especially in sparsely populated geographic regions where few genetics-trained providers are available, may result in patients having to travel longer distances to obtain recommended genetic services or not receiving them at all.[78] They also were concerned that patients seen by providers who were not trained in genetics could be poorly advised about testing options, inappropriately offered genetic testing, or tested without proper counseling on the ethical, legal, familial, and social complexities involved. Furthermore, untrained providers may be more prone to misinterpreting test results.

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[75] Other factors in addition to licensing may influence a provider's ability to participate in a health plan's provider network, including the provider's accessibility (e.g., scheduled office hours) to plan members, the health plan's provider needs, professional liability insurance, the sufficiency of health plan personnel, the system's ability to contract with and incorporate new providers, and, in the case of nonphysician providers, the availability of a contracted supervising physician.

[76] Cantrell M. Life after licensure: Our office's reimbursement experience. Presented at NSGC Annual Education Conference, October 2004.

[77] Public comments to SACGHS on draft Report on Coverage and Reimbursement of Genetic Tests and Services. June 2005.

[78] Ibid.

Recommendation 7

Genetic counseling is a critically important component of the appropriate use and integration of genetic tests and services.

7a To ensure full access to genetic counseling services for all Americans, the Secretary should expeditiously identify an appropriate entity to determine (1) which health professions are qualified to provide genetic counseling services (see page 49 for discussion of genetic counseling services and providers) and of those determined to be qualified, (2) which should be able to practice without physician supervision and, thereby, bill payers directly for their services. The entity selected to make these determinations should be guided by the professions' credentials, licensure status, scope of practice, and any other criteria deemed appropriate. The credentialing standards of a number of professional societies, such as the American Board of Genetic Counseling and the Genetic Nursing Credentialing Commission, could be used as a reference point. A description of existing credentialing programs is provided in Appendix B.

If this review process results in the determination that a health profession should be allowed to practice independently, the Secretary should urge Congress to add this health profession to the list of nonphysician practitioners eligible to bill Medicare directly for their services.

7b HHS should assess the adequacy of existing CPT E&M codes and their associated relative values with respect to genetic counseling services. This assessment should be carried out with input from genetic counseling service providers. HHS should address any inadequacies as deemed appropriate.

7c The Secretary should direct CMS to allow nonphysician health providers who are deemed qualified to provide genetic counseling services and who currently bill incident to a physician to use the full range of CPT E&M codes available for genetic counseling services.

7d The Secretary should ensure that all HHS programs are reimbursing prolonged service codes when they are determined to be reasonable and necessary.

7e The Secretary should direct CMS to deem all nonphysician health providers permitted to bill a health plan directly as eligible for a National Provider Identifier.

Broader Issues

Three other broad issues—health disparities, education and training of providers, and public awareness—warrant some discussion because of the effect they may have on the collection and dissemination of information to decisionmakers and, in turn, on coverage decisionmaking.

Health Disparities

Health care disparities according to gender, race/ethnicity, socioeconomic status, disability, geographic location, and sexual orientation have been well-documented in the United States and are attributed to a number of causes, including underutilization of health services in particular groups. For example, differences in the use of angioplasty have been documented by gender and race. When new tests are underutilized in some populations, data about the test's utility may be incomplete and inaccurate for those groups. In such instances, evidence may be deemed insufficient to justify coverage of that new genetic test or service for certain populations, which may further exacerbate existing health care disparities.

In the future, drug development based on pharmacogenomics may guide drug therapy decisions and improve the efficacy and safety of drug treatment. However, as more drugs are developed based on genotype, individuals with other genotypes may find treatment options to be limited or lacking, which could exacerbate current health care disparities (if genotype varies by gender, race/ethnicity, socioeconomic status, etc.). Health insurance plans are likely to be particularly challenged by pharmacogenomic-based drugs as they determine which drugs to cover and include on their list of preferred drugs. It may be difficult to find the right balance between providing access to the most effective drug based on genotype and providing access to drugs for the largest number of people.

Provider Education and Training

Increasingly, genetic tests are being marketed to health providers and directly to consumers. If providers are not adequately trained in the use and interpretation of genetic tests, they may provide inappropriate services to their patients and expect to be reimbursed for them. Health providers' use of, and thus demand for reimbursement for, new genetic tests will be affected by their education and training in genetics and genomics. Providers need adequate genetics education and training to help their patients make decisions about when to be tested. Genetics education and training should be a multidisciplinary and interdisciplinary process provided throughout the course of training so that health providers are able to judge the merits of new genetic tests as they appear.

A working knowledge of genetics also is important for health payers. These individuals, who are often physicians, need an understanding of genetics to make informed and appropriate coverage decisions.

Recommendation 8

Since genetic tests and services are being integrated into all areas of health care and since providers have an important role in ensuring appropriate use of and access to genetic tests and services among diverse populations, there is a critical need for programs to educate and train health providers and payers in genetics and genomics. Health providers should be able to meet established genetic competencies and, thereby, integrate genetics effectively into their practices. The Secretary should develop a plan for HHS agencies to work collaboratively with Federal, State, and private organizations to develop, catalog, and disseminate case studies and practice models that demonstrate the relevance of genetics and genomics.[1]

The Secretary should provide financial support to assess the impact of genetics education and training on health outcomes.

The Secretary should strive to incorporate genetics and genomics into relevant initiatives of HHS, including the National Health Information Infrastructure.

[1] SACGHS Resolution on Genetics Education and Training of Health Professionals, June 2004. http://www4.od.nih.gov/oba/sacghs/reports/EducationResolutionJune04.pdf [December 14, 2005].

Public Awareness

Public awareness of new health care tests and treatments can create consumer demand for them. The trends of less managed care and increasing utilization of prescription drugs demonstrate the influence of consumer demand on health care. Pressure from consumers on health care payers also can have an impact on coverage and reimbursement decisions.

Education can empower consumers in relation to payers. A decision influenced by consumer demand may be appropriate when this demand is based on valid and complete information. However, consumer demand also can be based on inaccurate and incomplete information. In these cases, consumers may demand coverage of services that have not been deemed to be safe or effective in certain circumstances. For example, consumer demand contributed to health insurance plans' decisions to cover autologous bone marrow transplants for breast cancer even though there were questions raised about the safety and efficacy of this treatment. Consumers also may demand coverage for services whose medical necessity is questionable (e.g., drugs for erectile dysfunction). Although providing such services may improve quality of life, its health value is less obvious, as is the obligation of a health insurance plan to provide coverage.

With respect to new genetic tests, public awareness and consumer demand could play an important role in facilitating coverage for new, safe, and effective interventions. Appropriate and timely coverage of new genetic tests could help facilitate and speed the broad translation of genetics into health care, thereby improving health outcomes, quality of care, and access to services. However, the complexity of genetics can result in misinformation and inappropriate demand for genetic tests and services. Media coverage has an important role in educating the public about genetic advances and communicating their relevance to health

and health care in a manner that is understandable to laypersons. Furthermore, basic genetics education at the K-12 level may provide a foundation for genetic literacy and public understanding.

Since genetics will have broad social impact, the role of public awareness and consumer demand with respect to coverage of genetic tests may be unique in relation to other medical tests. For this reason, consumers may have an important role in coverage decisions.

Recommendation 9

For patients and consumers to evaluate health plan benefits and health providers and make the most appropriate decisions for themselves and their families, they need reliable and trustworthy information about family history, genetics, and genetic technologies. The Secretary should ensure that educational resources are widely available through Federal Government Web sites and other appropriate public information mechanisms to inform decisions about genetic tests and services.

Appendix A

Health Care Financing in the United States

Health care in the United States is financed and delivered through a mixed public-private system. In this system, the Federal Government, State governments, employers, private health plans, providers, patients, and consumers all have a role in purchasing and providing health care and in making health care decisions. An understanding of the complexity of the U.S. health care system is needed to appreciate the limitations of the current system and the impact of these entities on coverage and reimbursement of genetic tests and services.

Unlike citizens of countries with government-sponsored universal health care systems, not all Americans have health insurance coverage. In 2003, 84 percent of Americans had some type of health insurance, either through a public program or a private health plan.[79] Sixteen percent were without any type of coverage.[80]

Government Programs[81]

Public health programs include any health services or health insurance administered or sponsored by the Federal or State Governments. The Federal Government is the largest sponsor of health care in the United States, providing health insurance and health services to elderly persons, disabled individuals, certain low-income groups, Federal Government employees, American Indians and Alaska Natives, veterans, and military personnel (active and retired) and their families. State governments also have an important role in providing health care and health insurance to State government employees and certain low-income groups and high-risk individuals.

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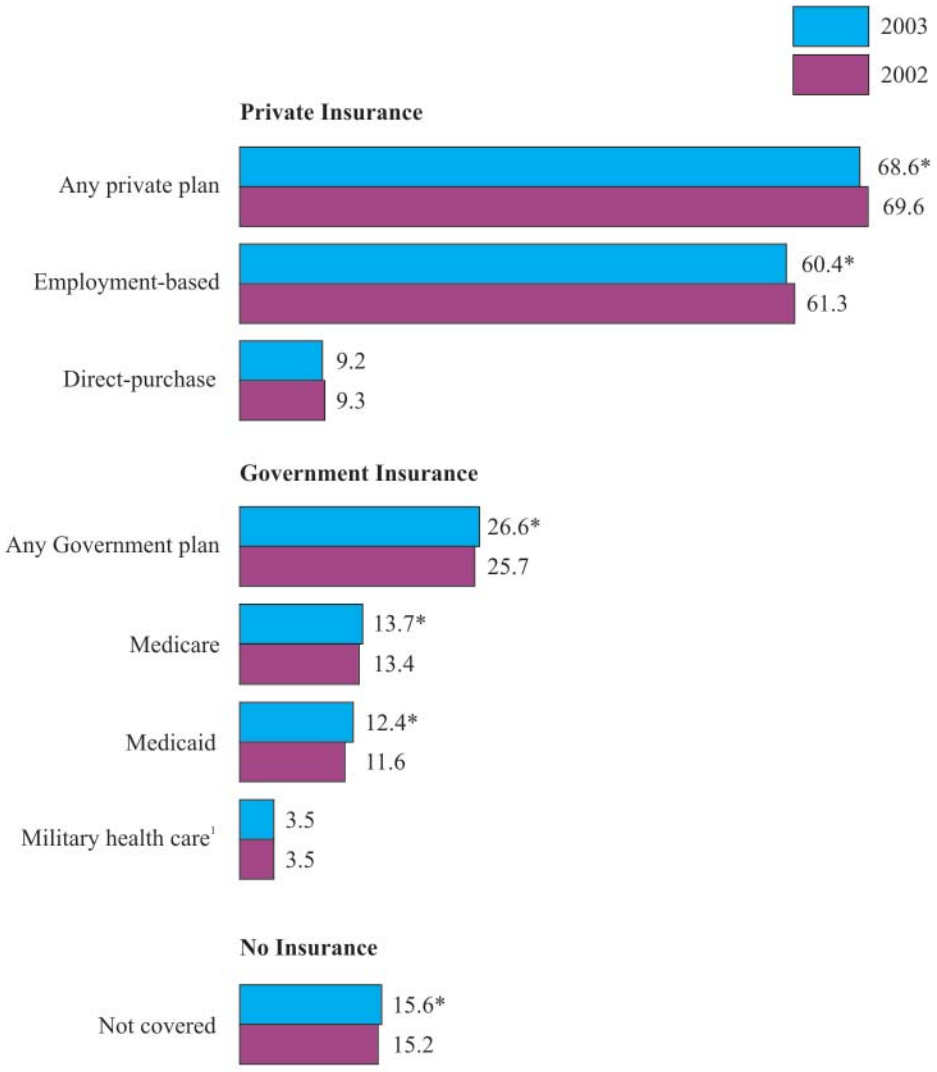
[79] Some individuals have both public and private health insurance (e.g., elderly persons with Medicare public insurance and Medigap private insurance. Thus, the percentages do not total 100 percent).
[80] U.S. Census Bureau. Income, Poverty, and Health Insurance Coverage in the United States: 2003. http://www.census.gov/prod/2004pubs/p60-226.pdf [November 18, 2005].
[81] This section is largely informed by the data and background materials from the Centers for Medicare & Medicaid Services and the Kaiser Family Foundation, unless specified otherwise.

Coverage by Type of Health Insurance, 2002 and 2003
(percent)



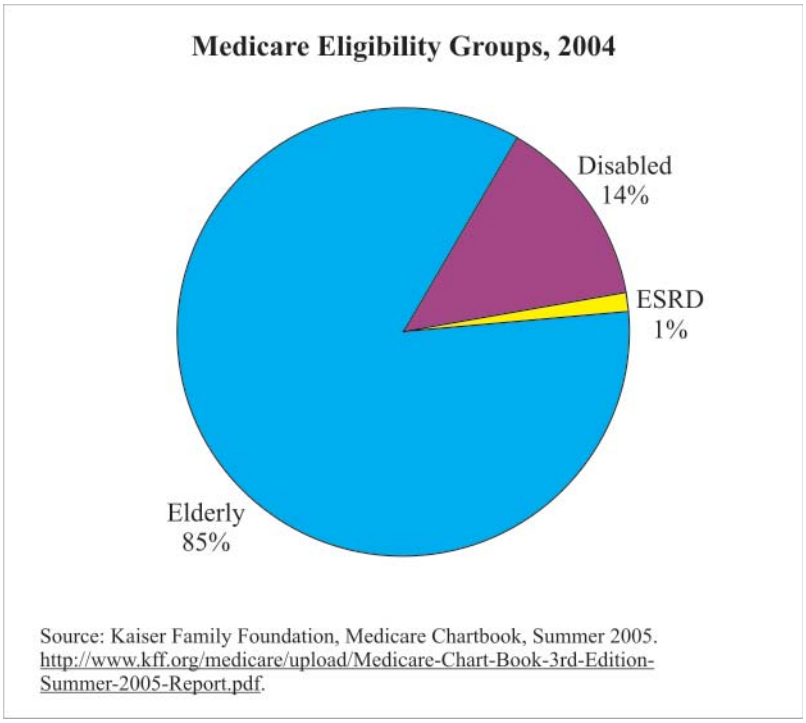
* Statistically different at the 90-percent confidence level.
[1] Military health care includes: CHAMPUS (Civilian Health and Medical Program of the Uniformed Services)/ Tricare and CHAMPVA (Civilian Health and Medical Program of the Department of Veterans Affairs), as well as care provided by the Department of Veterans Affairs and the military.

Note: The estimates by type of coverage are not mutually exclusive; people can be covered by more than one type of health insurance during the year.

Source: U.S. Census Bureau, Current Population Survey, 2003 and 2004 Annual Social and Economic Supplements.

Source: U.S. Census Bureau. Income, Poverty, and Health Insurance Coverage in the United States: 2003. http://www.census.gov/prod/2004pubs/p60-226.pdf [November 18, 2005].

Medicare



Source: Kaiser Family Foundation, Medicare Chartbook, Summer 2005. http://www.kff.org/medicare/upload/Medicare-Chart-Book-3rd-Edition-Summer-2005-Report.pdf.

Established in 1965 by the Social Security Act and administered by the Centers for Medicare & Medicaid Services (CMS), Medicare is a federally funded health insurance program that provides health benefits to more than 41 million Americans age 65 and older, or younger than age 65 with certain disabilities or end-stage renal disease (ESRD).

Covered benefits are divided into two parts: Part A for hospital insurance and Part B for medical insurance. Part A services include inpatient hospital, skilled nursing facilities, hospice, and some home health care. Part B includes physician services, outpatient hospital care, laboratory tests, medical supplies, and some home health care not covered under Part A. Prescription drug benefits were added in 2003 to begin in 2006. In general, Medicare does not cover preventive services unless explicitly mandated by Congress. Examples of preventive services whose coverage was recently authorized by Congress are an initial wellness physical examination and screening for diabetes and heart disease.

Twelve percent of Medicare beneficiaries have chosen to enroll in Medicare Advantage (previously known as Medicare+Choice) or the Medicare Cost Program (Medicare's managed care options) rather than to receive benefits under the Medicare fee-for-service program. Under these options, private health plans contract with Medicare to provide, at a minimum, Part A and B benefits to enrolled beneficiaries. Medicare Advantage plans typically cover additional benefits beyond those provided under Parts A and B. Currently, 247 private health plans have contracts to offer Medicare Advantage.[82]

Twenty-one percent of Medicare beneficiaries purchase supplemental insurance (known as "Medigap") from a private health plan to obtain coverage for health services not covered by Medicare. Also, some beneficiaries receive supplemental coverage from Medicaid (17 percent) or their previous employer (35 percent) as part of their retirement benefits.

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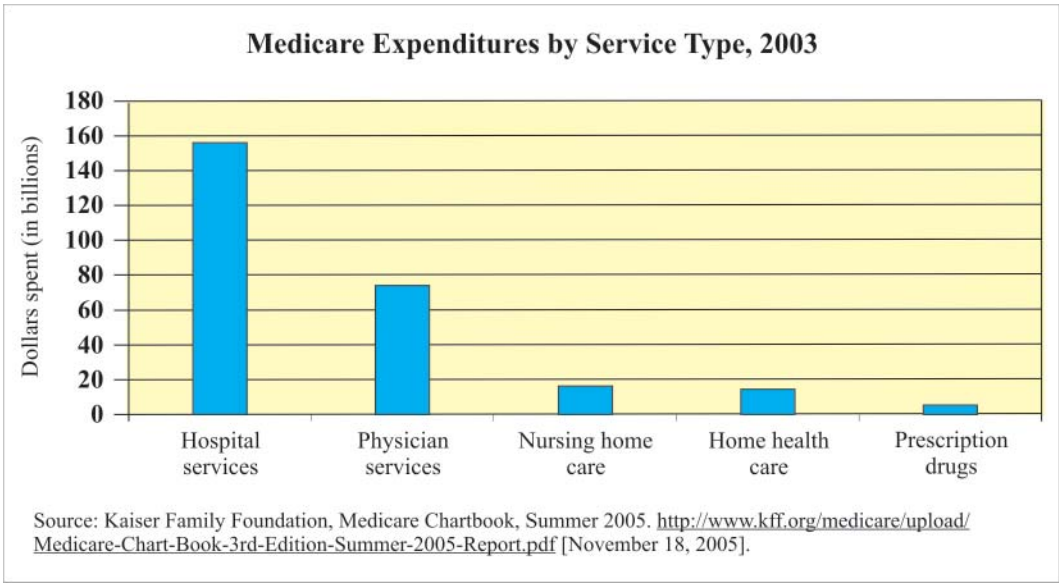
[82] Kaiser Family Foundation, Medicare Health and Prescription Drug Plans Monthly Tracking Report, July 2005. http://www.kff.org/medicare/upload/Medicare-Chart-Book-3rd-Edition-Summer-2005-Report.pdf [November 18, 2005].



Source: Kaiser Family Foundation, Medicare Chartbook, Summer 2005. http://www.kff.org/medicare/upload/Medicare-Chart-Book-3rd-Edition-Summer-2005-Report.pdf [November 18, 2005].

Medicaid and SCHIP

Also established in 1965 by the Social Security Act, Medicaid provides health insurance to over 52 million low-income Americans. Each State finances and administers its own Medicaid program and receives matching funds from the Federal Government. CMS oversees the Medicaid program at the national level.

The Federal Government has established minimum eligibility criteria for Medicaid; however, States have the discretion to broaden eligibility. At a minimum, a person must be (1) below a specified income level and (2) be younger than age 19 or age 65 and older, pregnant, blind, disabled, or have dependent children. In the case of low-income elderly persons, Medicaid will pay for Medicare premiums and for some services not covered by Medicare. Several States, such as Tennessee (TennCare) and Minnesota (MNCare), have received waivers from CMS allowing them to extend Medicaid coverage to individuals who are uninsurable for medical reasons or who have lost access to employer-subsidized health insurance and cannot afford private insurance.

Federal law requires State Medicaid programs to cover inpatient and outpatient hospital services; provider services; laboratory and x ray services; nursing home and home health care; early and periodic screening, diagnosis and treatment for children younger than age 21; family planning; pregnancy care; and rural health clinics and federally qualified health centers. States may cover other services in addition to these.

In an effort to contain costs and promote access and quality, many States have contracted with private managed care plans to provide health care coverage to Medicaid beneficiaries. In 2004, 61 percent of Medicaid beneficiaries were enrolled in Medicaid managed care plans.[83]

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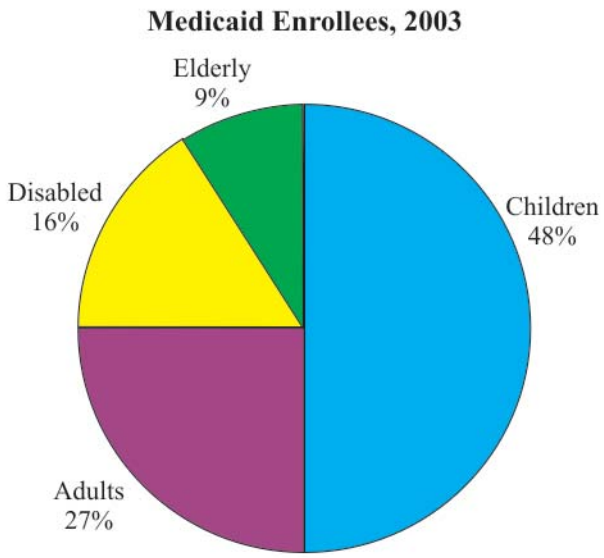
[83] CMS. 2004 Medicaid Managed Care Enrollment Report. http://new.cms.hhs.gov/MedicaidDataSourcesGenInfo/Downloads/mmcer04.pdf [January 4, 2006].

In 1997, Congress authorized $40 billion in matching funds to be given to States over a 10-year period to establish the State Children's Health Insurance Program (SCHIP). The program provides health insurance to uninsured children younger than age 19 with family incomes that are 200 percent of the Federal poverty level or 50 percent higher than the State's Medicaid income threshold but who are not eligible for Medicaid or are not covered by private insurance. Like Medicaid, SCHIP is a State-administered program, with each State given the flexibility to broaden eligibility and services. States have implemented SCHIP in various ways: some States expanded Medicaid, others opted to create separate programs, and still others have combined plans. Together, Medicaid and SCHIP account for 15 percent of U.S. health expenditures.



Source: Kaiser Family Foundation, Medicaid Program at a Glance, January 2005. http://www.kff.org/medicaid/upload/The-Medicaid-Program-at-a-Glance-Fact-Sheet.pdf [November 18, 2005].

Other Government Programs

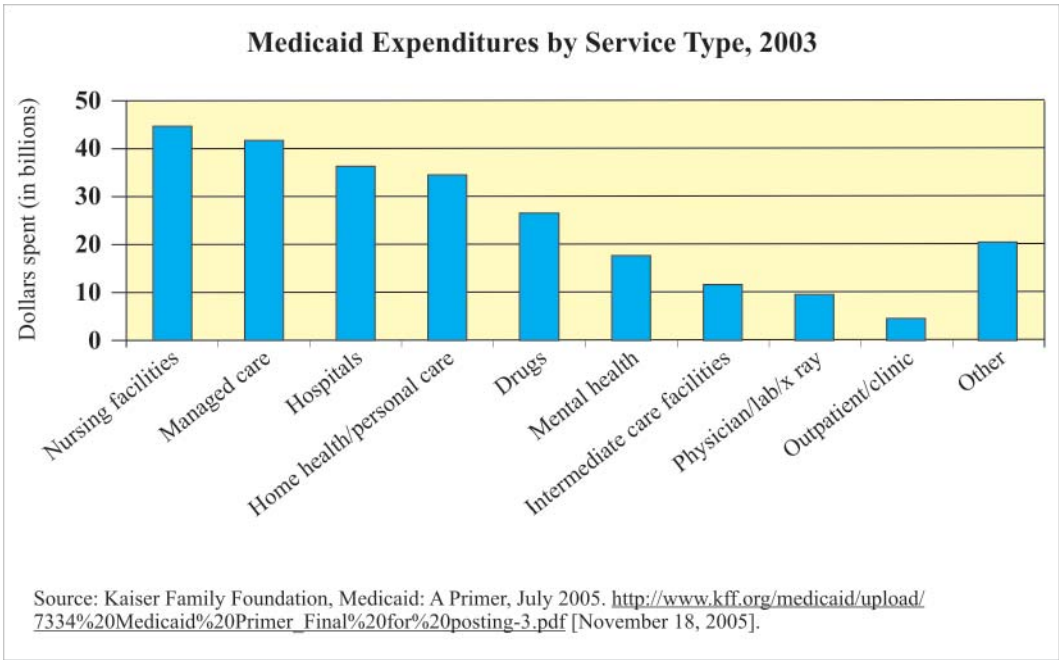
Federal Employees Health Benefits Program. Covering 9 million Federal employees, retirees, former employees, family members, and former spouses, the Federal Employees Health Benefits Program is the largest group health insurance program in the world. Under this program, the Federal Government pays up to 75 percent of the health plan premium. Over 350 plans currently participate in the program.

Veterans Health Administration. The Veterans Health Administration is the Nation's largest integrated health care system, providing primary, acute, specialized, rehabilitative, and long-term care and related medical and social support services to individuals honorably discharged or released from active duty service in the military service.

TRICARE. TRICARE (formerly known as CHAMPUS, the Civilian Health and Medical Program of the Uniformed Services), is the military managed care health system for active duty and retired members of the uniformed services as well as their families and survivors.

Indian Health Service. The Indian Health Service is a comprehensive health services delivery system operated by HHS for American Indians and Alaska Natives.

Federal Prison Health System. The Federal Bureau of Prisons' Health Programs Branch coordinates the medical, dental, and mental health services provided to Federal prison inmates.



Source: Kaiser Family Foundation, Medicaid: A Primer, July 2005. http://www.kff.org/medicaid/upload/7334%20Medicaid%20Primer_Final%20for%20posting-3.pdf [November 18, 2005].

State High-Risk Pools. Thirty-two States currently have high-risk insurance pools for individuals who were denied coverage, who were offered coverage with riders or exclusions or rates that exceed a determined amount (e.g., 120 percent of the individual market average), or with certain medical conditions (e.g., AIDS).[84]

Small Business Insurance Pools. Because of the few number of employees, small businesses can experience difficulty affording health insurance for their employees and, as a result, are less able to provide health benefits to their employees compared with large companies. To improve small businesses' purchasing power, 15 States sponsor purchase alliances for small businesses.

State and County Plans. State and county governments also offer health insurance plans to employees and their families.

Private Health Insurance Plans

Seventy percent of health insurance provided in the United States is purchased through a group (such as one's employer, union, or professional association) or directly from a health insurance plan, health maintenance organization (HMO) or preferred provider organization (PPO) on an individual basis. Many employers offer several different types of plans, allowing employees to select the health insurance plan that is most affordable and best suits their and their families' health care needs.

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[84] State High-Risk Health Insurance Pool Participation, December 31, 2003. http://statehealthfacts.org/cgi-bin/healthfacts.cgi?action=compare&category=Managed+Care+%26+Health+Insurance&subcategory=High+Risk+Pools&topic=High+Risk+Pool+Participation&link_category=&link_subcategory=&link_topic=&printerfriendly=0&viewas=table [December 5, 2005].

Group Market. Many large employers and small businesses offer health benefits to their employees and employees' family members, in part due to the availability of tax incentives. In most employer-sponsored health plans, the employer pays a portion of the premium, and the balance is deducted from the employee's pay. Because employers pay for a considerable portion of health care costs in the United States, they have a significant role in coverage and reimbursement decisions.

Some large employers are able to assume the financial risk for their employees' medical costs. These self-insured employers may administer their health plan(s) in-house or contract with a third-party administrator to administer benefits. The Employee Retirement Income Security Act (ERISA) generally preempts any State law that relates to employee benefit plans,[85] with the exception of State laws that regulate insurance. As a result, self-insured employee benefit plans are generally only subject to ERISA, whereas insured employee benefit plans must comply with both ERISA and any State insurance regulations, including mandated benefits laws and premium taxes.

Other employers transfer the risk for medical costs through a contract with a health insurance issuer. As with Medicare managed care, these employers pay private health insurance plans to assume responsibility for their employees' medical costs and to administer benefits. Because health insurance is purchased as a group, because the group was not formed for the purpose of purchasing health insurance, and because enrollment is permitted only at the beginning of eligibility or during open enrollment periods, group coverage is less risky for the health insurance company than coverage purchased by an individual. Thus, health insurance companies are less likely to scrutinize the medical histories of individual employees and their potential likelihood of needing future health care as a condition for group coverage.

What is the purpose of health insurance?

In principle, health insurance allows individuals to transfer the risk of unpredictable costs to a third party for a predetermined price, known as a premium. In exchange for this premium, the third party accepts responsibility for paying for any health services covered by the health insurance contract.

Health insurance began as a way for individuals to protect themselves from catastrophic medical costs. Over time, however, health insurance has metamorphosed into a more comprehensive product that covers not only costly, unforeseeable, lifesaving medical care but also other services whose costs are more predictable and have downstream, uncertain, or questionable impact on a person's

Individual Market. Individuals also may purchase health insurance directly from a private health insurance company. Nine percent of Americans obtain health insurance this way. Unlike group coverage, individual coverage purchased directly from a health insurance company often requires disclosure of family and personal health history and current medical problems to assess whether the individual is insurable from an actuarial standpoint, to determine whether any services should be excluded from the policy, and to set an appropriate premium rate based on the person's risk classification.

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[85] An employee benefit plan (EBP) is a legal entity called a "plan" that is created when employers or employee organizations, usually joint labor/management boards sponsoring collectively bargained plans, voluntarily enter into arrangements to provide benefits such as pensions, vacation, day care, prepaid legal services, or medical care through insurance or otherwise. EBPs can be either pension plans or "welfare benefit plans."

Plan Types

Employers and individuals have a range of health plan products to choose from. In general, health plans are classified as indemnity or managed care. Although not as popular, consumer-driven health plans have been gaining attention in recent years.

Indemnity Plans

When private health insurance first began, it took the form of indem-nity insurance. Today, indemnity insurance accounts for only 5 percent of health insurance enrollment. Indemnity insurance plans are distinct from managed care because they allow unrestricted access to any licensed provider covered under the plan and reimburse providers for medically necessary services on a fee-for-service basis.[86] Many indemnity plans have adopted management features used by managed care plans, blurring the distinction between the two categories. Indemnity plans cover preventive services less frequently than managed care plans. Because there are fewer constraints on health care utilization with indemnity insurance, premiums tend to be higher than with managed care, and as a result, these plans are less prevalent.

Network-Based Plans

In response to escalating health care costs in the 1970s and 1980s, managed care plans became a popular alternative to indemnity plans among health care payers for their ability to better control health care utilization and costs. Several variations on traditional managed care plans have developed over the years. These plans are all network based and have retained certain, but not all, of the characteristics of traditional managed care (e.g., preauthorization and physicians acting in a gatekeeper capacity). Today, network-based plans account for 85 percent of the health insurance market. These plans are characterized by their dual role in arranging for and financing health care. They do this by entering into contracts with providers to service their members for negotiated rates and by providing incentives to enrollees to utilize contracted providers.

Health Maintenance Organizations. Health maintenance organizations (HMOs) not only are considered the most managed of the network-based plans but also often have the lowest premiums. HMOs contract with health providers to supply care to their plan members. HMOs that are "open" usually allow their providers also to see non-HMO patients or patients from other HMOs. Providers participating in "closed" HMOs (i.e., staff model) generally see only patients enrolled in the particular HMO. Such staff-model HMOs are no longer very prevalent, with Kaiser Permanente perhaps the most well-known example still in existence. HMO-provider contracts typically require providers to bear some financial risk through capitated payments and provide incentives to providers and members to limit utilization of unnecessary services in an effort to control costs.

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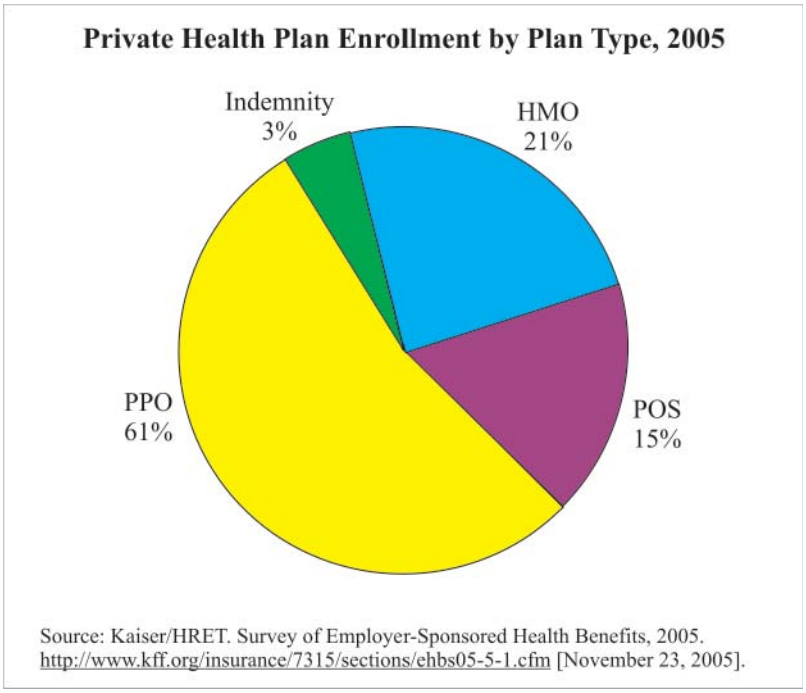
[86] Fee-for-service refers to a payment method in which the health plan reimburses the provider retrospectively for services provided. In contrast, many managed care plans utilize prospective payment for at least some of their contracted providers (typically primary care providers and some specialists). With prospective capitated payment, providers are paid a predetermined, per-member-per-month fee regardless of whether any services are provided to the member. Providers are allowed to keep any overpayment but must accept the loss for underpayment under a capitated payment system. This method provides incentive to providers to provide only necessary care.

Members often are required to select a primary care provider and obtain a referral for an in-network provider in order for the service to be covered by their HMO. Services obtained from out-of-network providers with whom their HMO does not have a contract are usually not covered.



Point of Service Plans. Point of service (POS) plans are similar to HMOs but provide members with the option at the point of care to see an in-network provider or pay a greater share of the cost to see an out-of-network licensed provider covered under the POS plan.

Preferred Provider Organizations. Preferred provider organizations (PPOs) are the most flexible and most popular of the managed care plan types, with 54 percent of individuals with private health insurance enrolled in PPOs. As with POS plans, members have the option to see an in-network provider or pay a greater share of the cost to see an out-of-network licensed provider covered under the PPO plan. Unlike HMOs and POSs, in-network providers are usually paid on a fee-for-service basis at a negotiated discounted rate and do not share any financial risk with the PPO.

Consumer-Driven Health Plans

Consumer-driven health plans (CDHPs) have generated much interest in recent years as a way to control health insurance costs by providing individuals with increased information on quality for enhanced decisionmaking and choice and control of their health benefits and expenditures. It is still too soon to say whether these new health plan types will become part of the mainstream; however, they may provide a better way to obtain services such as genetic tests and services that otherwise may not be offered by more traditional health plans. If CDHPs gain popularity, the particular impact they might have on the integration of genetic tests and services and any unique challenges they may raise will need to be assessed.

Health Savings Accounts. A health savings account is a tax-advantaged account that can be funded with employer and/or employee dollars. These accounts can be used only for qualified medical expenses (or the tax advantages may be lost), and they must be paired with a high-deductible health plan. The accounts are owned by the employee and, thus, are fully portable. Any balance remaining at the end of the year can be carried over.

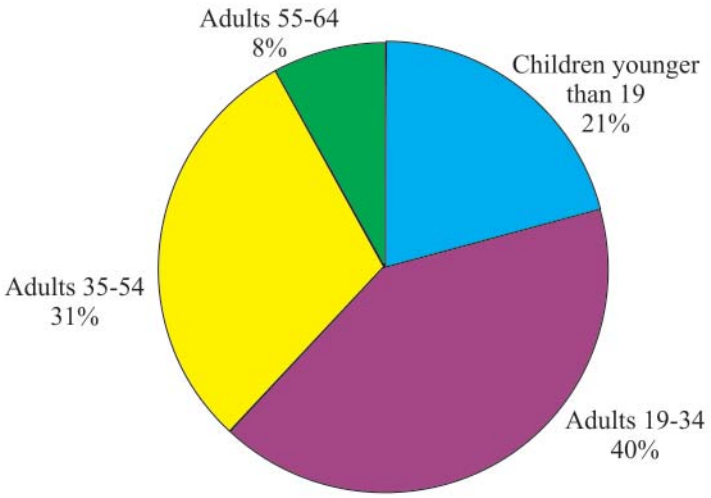
Health Reimbursement Arrangements. A health reimbursement arrangement (HRA) is another tax-advantaged way for employers to provide coverage for medical benefits. In an HRA, the employer reimburses employees for qualified medical expenses up to a predetermined limit. HRAs may, but are not required to,

be paired with other health coverage. Any unused allotment remaining at the end of the year can be carried over.

Personalized Plans. Personalized or design-your-own plans allow members to use an Internet-based tool to create their own provider networks and benefits packages, with the members' selections determining the cost of the plan. The member is responsible for any costs that exceed the employer's fixed contribution.

Customized Package Plans. In customized package plans, the member chooses from among several predetermined network types and benefits plans. As with personalized, design-your-own plans, the member is responsible for any costs above the defined employer contribution.

Characteristics of Uninsured Individuals by Age, 2002



Source: Kaiser Commission on Medicaid and the Uninsured. The Uninsured: A Primer- Key Facts about Americans without Health Insurance. http://www.kff.org/uninsured/upload/The-Uninsured-A-Primer-Key-Facts-about-Americans-without-Health-Insurance.pdf [November 18, 2005].

Uninsured and Underinsured Individuals

Although this report does not address problems in access to genetic tests and services resulting from a lack of health insurance, it is important to acknowledge that 43.6 million Americans were uninsured in 2002 (15 percent of the population). This group is largely made up of working families, young healthy adults, immigrants, and individuals with low income or in poor health. Uninsured rates vary by geographic region and race/ethnicity.

Although not as easy to characterize, underinsurance is also a significant problem in the U.S. health care system. Underinsured persons have health insurance but are responsible for a significant amount of out-of-pocket costs, and/or their coverage is limited in scope or amount. Individuals who purchase insurance individually (i.e., not through their employer) or who work for small businesses are most prone to being underinsured.

As demonstrated, there are a number of models for organizing the financing of health care. As such, policies that increase access to genetic tests and services will need to be relevant to a variety of public and private purchasers and multiple arrangements of health providers, and increasingly responsive to individual consumers who will be responsible for making health decisions regarding their benefits and use of health care resources.

Appendix B

Report of the Work Group on Genetic Counseling Services

This report was prepared by an independent work group formed to respond to a request for evidence on topics relevant to the coverage and reimbursement of genetic counseling services to inform the Committee's deliberations in this area. It does not represent the official views of SACGHS or the U.S. Government.

This report, produced in February 2005, was developed by a work group enacted by the SACGHS staff in November 2004. Work group members include Judith Cooksey (workforce issues), Andrew Faucett and Anne Greb (American Board of Genetic Counseling or ABGC), Dale Lea (International Society of Nurses in Genetics or ISONG), and Kelly Ormond (National Society of Genetic Counselors or NSGC). Daniel Riconda, a genetic counselor with expertise in genetic counseling licensure, current chair of the NSGC licensure subcommittee and ABGC board member, also was a work group member. SACGHS member Cynthia Berry served as an adviser to the work group.

SACGHS asked the Work Group on Genetic Counseling Services to consider the following issues related to billing and reimbursement for genetic counseling services: (1) the credentials and qualifications of various nonphysician genetic service providers (including how it relates to the licensure of genetic counselors); (2) the value and effectiveness of genetic counseling services; and (3) the importance of reimbursing genetic counseling services, benefits in doing so, and potential harms if reimbursement is not obtained.

Introduction

As has been previously presented to SACGHS, health professionals who are specifically trained and dedicated to the provision of genetic counseling and clinical genetic services include master's level genetic counselors; genetic nurses trained at the bachelor's, master's, and PhD levels; MD clinical geneticists; and PhD medical geneticists.[87] There currently are 1,178 MD clinical geneticists who are board certified by the American Board of Medical Genetics (ABMG) and 152 ABMG-board certified PhD medical geneticists.[88] ABGC reports 1,811 ABMG/ABGC-board certified genetic counselors, with an additional 466 eligible for the 2005 examination cycle. ISONG reports 39 individuals credentialed as either an advanced practice

[87] Judith Cooksey, Preliminary findings of the genetics workforce study, presentation to SACGHS October 23, 2003 meeting. http://www4.od.nih.gov/oba/SACGHS/meetings/October2003/Cooksey_tr.pdf [November 25, 2005].
[88] ABMG. Number of certified specialists in genetics. http://www.abmg.org/genetics/abmg/stats-allyears.htm [December 9, 2005].

nurse in genetics (APNG) or a genetic clinical nurse (GCN). Thirty ISONG nurses are also board certified in genetic counseling. Overall, there are currently 3,076 professionals in the United States who are specifically trained, certified, and dedicated to provide genetic counseling and clinical genetic services, of which 59 percent are master's-level genetic counselors. The number of physicians presenting for clinical genetics training (in either residency or fellowship) has been declining since 1996 according to data presented at the Banbury Summit on Genetics Training held in October 2004.[89] In contrast, the number of genetic counselors trained increase steadily during the same time period.

Training, Qualifications, and Credentials of Providers of Genetic Counseling Services

Coverage and reimbursement depend on the qualifications of the providers. This section details the credentials and qualifications of genetics professionals.

The genetic counseling process involves the collection and interpretation of family, genetic, medical, and psychosocial history information. Analysis of this information, together with an understanding of genetic principles and the knowledge of current technologies, provides patients and their families with information about risk, prognosis, medical management, and diagnostic and prevention options. Information is discussed in a client-centered manner while respecting the broad spectrum of beliefs and value systems that exist in our society. The genetic counseling process ultimately facilitates informed patient decisionmaking and promotes behaviors that reduce the risk of disease. As described later in this report, the training needed to competently provide genetic counseling is specialized and includes coursework and hands-on supervised clinical experiences. This combination of coursework and clinical training distinguishes these individuals from other health providers who may occasionally provide genetic information.

Medical groups and organizations are beginning to recognize the importance of ensuring that qualified health professionals provide genetic counseling services. The National Cancer Institute has established a Cancer Genetics Services Directory that includes only the names of health providers who have met specified criteria and therefore are felt to be competent providers of genetic services.[90] Another example is the Minimum Guidelines for the Delivery of Prenatal Genetics Services, published by the Great Lakes Regional Genetics Group. These recommendations specify that "an ABMG-certified or eligible MD or PhD clinical geneticist or an ABGC-certified or board-eligible genetic counselor is available for consultation or case review" and that noncertified individuals should be supervised by a geneticist or genetic counselor.[91]

Training and Credentialing for Master's-Level Genetic Counselors. In 1969 the first graduate program to specifically train genetic counselors to provide genetic counseling was established at Sarah Lawrence College. Training program guidelines were subsequently established and included course work in counseling theories and techniques, human and medical genetics, molecular biology, and genetic counseling. Extensive supervised clinical training is considered an important component of genetic counselor training.[92]

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[89] Korf BR et al. Report of Banbury Summit meeting on training of physicians in medical genetics, October 20-22, 2004. *Genet Med* 2005. 7(6):433-8.

[90] National Cancer Institute. Cancer Genetics Services Directory: Criteria for Inclusion. http://www.cancer.gov/forms/joinGeneticsDirectory [November 25, 2005].

[91] Sommer A et al. Minimum guidelines for the delivery of prenatal genetics services. The evaluation of clinical services subcommittee, Great Lakes Regional Genetics Group. *Genet Med* 1999. 1(5):233-4.

[92] Walker AP et al. Report of the 1989 Asilomar Meeting on Education in Genetic Counseling. *Am J Hum Genet* 1990;46:1223-30.

ABMG was established in 1981 to certify MD and PhD geneticists, master's-trained genetic counselors, and nurses with a master's degree and concentrated training in genetics. Eligibility for genetic counselor certification includes a master's degree in a relevant discipline and a logbook of 50 cases obtained at approved clinical sites. ABMG originally certified 631 genetic counselors.

In 1992 the American Board of Medical Specialties recognized ABMG, and as a result, genetics residency programs were established and accredited by the Accreditation Council for Graduate Medical Education. ABGC incorporated in 1993 and took over the certification process for master's-level genetic counselors. Some 495 individuals with ABMG certification became charter members of ABGC. Individuals who did not meet the eligibility criteria to take the certification exam could apply to do so under special considerations through the 1999 exam. This mechanism allowed health professionals such as nurses and social workers who were experienced in providing genetic counseling but had nontraditional training to become certified by ABGC. The credentialing of PhD-trained medical geneticists remained with ABMG.

ABGC also began, for the first time, accrediting genetic counseling graduate programs. Practice-based competencies that define the role of a genetic counselor were established.[93] They include four domains: (1) communication skills; (2) critical-thinking skills; (3) interpersonal, counseling, and psychosocial assessment skills; and (4) professional ethics and values. The accreditation criteria for training programs were based on the program's ability to develop these competencies in its graduates. Although each program's curriculum and method of supporting the development of the practice-based competencies are unique, programs must provide instruction in the following general content areas: (1) principles of human, medical, and clinical genetics; (2) psychosocial theory and techniques; (3) social, ethical, and legal issues; (4) health care delivery systems and principles of public health; and (5) teaching techniques and research methods.

The clinical training must provide students hands-on experience working with individuals and families affected with a broad range of genetic disorders. These supervised experiences must expose students to the natural history, management, and psychosocial issues associated with common genetic conditions and birth defects. Students must have opportunities to develop their genetic counseling skills in a variety of clinical genetics settings.

Currently, the eligibility criteria to sit for the ABGC certification exam include:

- Graduation from an accredited genetic counseling graduate program.
- Fifty logbook cases acquired at approved sites. Cases must represent a wide variety of counseling roles and clinical situations and be supervised by ABMG- or ABGC-certified individuals. The applicant's role in each case must be clearly documented.
- Letters of reference from a program director and two board-certified genetics professional.

The general genetics examination used by both ABMG and ABGC is developed by ABMG, with ABGC contributing 10 percent of the questions. ABGC develops the specialty exam for genetic counseling, and ABMG develops specialty examinations for MD- and PhD-trained individuals. The National Board of Medical Examiners (NBME) is involved in the development and administration of the examinations for both

[93] Fiddler MB et al. A case-based approach to the development of practiced-based competencies for accreditation of and training in graduate programs in genetic counseling. *J Genet Counsel* 1996. 5:105-12.

boards. To date 1,675 additional genetic counselors have been certified by ABGC since 1993. Historically, the percentile scores on the general genetics examination for MD clinical geneticists and master's-level genetic counselors have not differed significantly, which further demonstrates genetic counselors' broad training in all areas of medical genetics.

To ensure that knowledge and skills are maintained, beginning in 1996, genetic counselors certified by ABGC are required to go through a recertification process every 10 years. Recertification can be obtained by reexamination or continuing education (25 hours/year).

Whereas ABGC provides for credentialing for the genetic counseling profession, NSGC is the national professional organization for genetic counselors. NSGC is the leading voice, authority, and advocate for the genetic counseling profession. NSGC has approximately 2,100 genetic counselor members, of whom 85 percent are certified by ABGC. In addition to providing many continuing education opportunities, NSGC has a code of ethics, produces position statements and practice guidelines, and is currently in the process of refining the definition of genetic counseling and a scope of practice for master's-level genetic counselors.

Credentialing for Genetics Nursing Practice. ISONG is an international nursing specialty organization that fosters the scientific and professional growth of nurses in human genetics. Incorporated in 1988, ISONG provides education and support for nurses providing genetics health care. ISONG promotes the integration of the nursing process into the delivery of genetic services and encourages the incorporation of the principles of human genetic principles into all levels of nursing education. As a professional society, ISONG establishes and maintains standards of practice for nurses in human genetics and supports advances in nursing research in human genetics.

ISONG has taken the lead in working with nursing leaders to promote genetic nursing practice and develop a credentialing process for genetic nurses. In 1997 genetics nursing was recognized by the American Nurses Association as an official specialty of nursing practice. In 1998 ISONG established the Scope and Standards of Genetics Nursing Practice for genetic nursing to ensure minimal levels of competency; these are currently being revised, with publication expected in 2005.

In 2001 ISONG approved formation of the Genetic Nursing Credentialing Commission (GNCC), which provides recognition for clinical nursing practice in health care with a genetics component. Beginning in 2001 nurses who are prepared with the master's in nursing may qualify for the Advanced Practice Nurse in Genetics (APNG) credential. APNG credentialing is based on submitting a portfolio of accomplishments documenting the nurse's genetic expertise. This includes:

- Registered Nurse (RN) with at least a master's degree in nursing or the equivalent
- At least 3 years of experience as a genetic nurse with a 50 percent genetic practice component
- Documentation of 50 cases where the APNG has provided health care services with a genetic component in the past 5 years
- Minimum of 50 contact hours of genetic content (e.g., acquisition of genetics content through classes, workshops, or continuing education) in the past 5 years
- Demonstration of clinical competency by submitting four indepth genetic case histories that show the nurse's ability to apply genetic knowledge according to the scope and standards of genetic nursing practice

- Submission of portfolio (including all of the above) that demonstrates the nurse's accomplishments and competency
- Three professional letters of reference

In 2002 GNCC was incorporated, and those prepared with a bachelor's degree in nursing may qualify for the Genetic Clinical Nurse (GCN) credential (first offered in 2002). The GCN credential also is by portfolio and for non-master's-prepared nurses.

Other nursing subspecialty organizations such as Oncology Nursing Society (ONS) and the Association of Women's Health Obstetrical and Neonatal Nurses (AWHONN) have established standards of practice and position statements, educational resources, and minimal practice requirements specific to their field for nurses practicing in genetics. The standards are consistent with the ISONG Standards of Practice but incorporate specialty-specific practice requirements. In addition, according to the National Coalition for Health Professional Education in Genetics (NCHPEG), there are 11 certified nursing specialties that incorporate genetics into their credentialing exam or core competencies. Finally, there are 11 graduate or graduate certificate programs that focus on genetics as well as 16 short courses and Web-based programs.[94]

Licensure of Nonphysician Genetic Service Providers. All health care providers—physicians, nurses, genetic counselors, mental health professionals, social workers, and other allied health professionals—assess risks and educate and inform patients. What distinguishes genetic counseling from education and counseling in other arenas is the combination of distinct and specific knowledge about genetics, inheritance, and human behavior (e.g., decisionmaking styles, coping mechanisms) combined with a focus on promoting autonomous decision-making through comprehensive informed consent. Therefore, the ability to provide genetic counseling requires a knowledge base and skill set that is distinct from other health professionals. The family's ability to make informed decisions about genetic testing, medical management, and lifestyle depends on the qualifications and competence of the health professional providing genetic counseling services.

As genetic health care moves into mainstream medicine, primary care providers and nongenetic specialists will be required to provide increasing genetic care. Several recent studies document that few primary care providers feel knowledgeable about genetics and genomics, and in many cases they are not comfortable providing many of the components of genetic counseling.[95,96] A number of additional peer-reviewed articles demonstrate the lack of adequate training of health care providers in genetic counseling.[97] Additionally, two court cases—Pate v. Threlkel (Florida) and Safer v. Pack (New Jersey)—address the consequences that can occur when health providers fail to recognize the familial nature of genetic conditions and the concomitant duty to warn relatives at risk. When one considers this in combination with the relatively small number of physician genetic specialists (approximately 1,100 nationally), it seems likely that current and future genetic

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[94] ISONG. 2005 Genetics and Health Workforce Survey Report.
[95] Pichert G et al. Swiss primary care physicians' knowledge, attitudes and perception towards genetic testing for hereditary breast cancer. *Fam Cancer* 2003. 2(3-4):153-8.
[96] Kussman J et al. Current and desired roles in the provision of genetic services among family physicians in the United States. *J Genet Couns* 2004. 13(6):543-4.
[97] Giardiello FM et al. The use and interpretation of commercial APC gene testing for familial adenomatous polyposis. *NEJM* 1997. 336(12):823-7. Other examples are listed in the bibliography at the end of Appendix B.

health care will require that services be provided by nonphysician genetic specialists, including nurses and genetic counselors with expertise in these areas. As such, ensuring the competence of our genetic workforce will be critically important.

Genetic counselor licensure, which is conducted on a State-by-State basis, creates specified standards for all health professionals providing genetic counseling services in that particular State. Genetic counselor licensure enables employers and the public in the State to know that practicing genetic counselors have achieved a minimal standard similar to licensure of physicians, nurses, and social workers. Additionally, most licensed health providers are required to maintain their continuing education units regardless of certification status. As a result of licensure, title protection may prohibit unqualified practitioners from being able to represent themselves as genetic counselors. If a consumer of genetic counseling services wishes to lodge a complaint, the Department of Health and the Department of Medical Quality Assurance (or Department of Professional Licensure or other authority in the State) will not record complaints unless licensure of the profession exists.

Nurses and MD geneticists working in genetics are already required to obtain State-based licensure in all States. Master's- and PhD-trained genetic counselors do not yet have licensure in most States. Utah is the only State that has currently enacted licensure of genetic counselors.[98] California and Illinois have passed legislation but not yet enacted it. Approximately 11 other States (Colorado, Florida, Massachusetts, New York, New Jersey, Oklahoma, Pennsylvania, Tennessee, Texas, Washington, and Wisconsin) currently have bills in the legislature or are drafting bills.

Some nongenetics professional or legal groups have explicitly supported the concept of genetic counselor licensure. The New York State Task Force on Life and the Law recommended the "state certification and promotion of genetic counseling as a profession,"[99] and the New York State District II American College of Obstetricians and Gynecologists (ACOG) chapter supports genetic counselor licensure.[100]

Licensure must be justified by demonstrating that consumers of genetic counseling services have been harmed by the lack of licensure regulations. Although anecdotal cases exist, published studies are lacking that document harm to consumers through the current lack of genetic counseling licensure; this may or may not be impacted by the inability for State departments to record complaints for a profession that is not yet licensed. State regulators may choose not to regulate genetic counselors if practitioners could be adversely affected, the cost to consumers is increased, the cost to professionals being licensed would be extreme (e.g., in a State with few numbers of practicing genetic counselors), or if access to such providers would be restricted or reduced as a result of such regulations.

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[98] Utah Administrative Code. Genetic Counselors Licensing Act Rules. www.rules.utah.gov/publicat/code/r156/r156-75.htm [November 25, 2005].
[99] New York State Task Force on Life and the Law. Genetic Testing and Screening in the Age of Genomic Medicine. 2001. http://www.health.state.ny.us/press/releases/2001/genetics.htm [November 25, 2005].
[100] American College of Obstetricians and Gynecologists. 2005 Legislative Program. www.acog.org/acog_districts/dist_notice.cfm?recno=1&bulletin=1508#_Toc84671574 [November 25, 2005].

The Value and Effectiveness of Genetic Counseling Services

Many professional practice standards (e.g., from organizations such as the American College of Medical Genetics, the American Society of Human Genetics, and ACOG) include the provision of genetic counseling services. Practice standards including genetic counseling services address reproductive genetic counseling services (e.g., around gamete donation, maternal serum screening, and the provision of prenatal diagnosis); carrier screening; and predictive testing (e.g., for breast/ovarian cancer). Some example statements include:

- "ASCO [American Society of Clinical Oncology] supports efforts to ensure that all individuals at significantly increased risk of hereditary cancer have access to appropriate genetic counseling, testing, screening, surveillance, and all related medical and surgical interventions, which should be covered without penalty by public and private third-party payers."[101]
- With regard to male infertility, "genetic counseling may be offered when a genetic abnormality is suspected in the male or female partner, and it should be provided when a genetic abnormality is detected."[102]
- "…such diagnostic and prenatal mutation analyses [for cystic fibrosis] should be referred to a genetics center for appropriate testing and counseling."[103]
- "All positive [Fragile X] results should state that genetic counseling is indicated… for at risk family members."[104]
- "Genetic counseling is provided whenever a prenatal testing procedure is performed"[105]
- "An essential component of a screening program is follow-up evaluation and counseling by genetic professionals for participants with positive results in order to assure appropriate understanding and treatment, and to reduce anxiety and stigmatization."[106]
- "Prospective couples should receive individualized genetic counseling [for advanced paternal age] to address specific concerns"[107]
- Regarding genetic research results, "it is strongly recommended that research results only be transmitted to subjects by persons able to provide genetic counseling."[108]

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[101] American Society of Clinical Oncology Policy Statement Update: Genetic Testing for Cancer Susceptibility. *J Clin Oncol* 2003. 21(12): 1-10. http://www.asco.org/asco/downloads/GeneticTesting.pdf [November 25, 2005].

[102] American Urological Association and American Society for Reproductive Medicine. Report on Optimal Evaluation of the Infertile Male. http://www.auanet.org/timssnet/products/guidelines/main_reports/optimalevaluation.pdf [November 25, 2005].

[103] Grody WW et al. Laboratory standards and guidelines for population-based cystic fibrosis carrier screening. *Genet Med* 2001. 23(2):149-54. http://www.acmg.net/resources/policies/pol-005.asp [November 25, 2005].

[104] Maddelena A et al. Technical standards and guidelines for Fragile X: The first of a series of disease-specific supplements to the standards and guidelines for clinical genetics laboratories of the American College of Medical Genetics. *Genet Med* 2001. 3(3):200-5. http://www.acmg.net/resources/policies/pol-013.pdf [November 25, 2005].

[105] Sommer A et al. Minimum guidelines for the delivery of prenatal genetics services. The evaluation of clinical services subcommittee, Great Lakes Regional Genetics Group. *Genet Med* 1999. 1(5):233-4.

[106] ACMG. Principles of screening: Report of the Subcommittee on Screening of the American College of Medical Genetics Clinical Practice Committee. http://www.acmg.net/resources/policies/pol-026.asp [November 25, 2005].

[107] ACMG. Statement on guidance for genetic counseling in advanced paternal age. http://www.acmg.net/resources/policies/pol-016.asp [November 25, 2005].

[108] ASHG. Statement on informed consent for genetic research. http://www.ashg.org/genetics/ashg/pubs/policy/pol-25.htm [November 25, 2005].

- "Women with a previous history of NTDs [neural tube defects] in their families should obtain genetic counseling concerning…"[109]

Massachusetts State law requires that all genetic testing be accompanied by a statement that the person was informed about the availability and importance of genetic counseling and provided with written information identifying a genetic counselor or medical geneticist from whom the consenting person might obtain such counseling.[110] Michigan and New York are among States specifying that informed consent must be obtained for specific genetic tests but do not specify who should provide such consent.[111]

Before reviewing the available evidence with regard to documenting the value and effectiveness of genetic counseling, it is critical to understand that such studies are difficult to interpret, for several reasons. First, studies assess genetic counseling services provided by physicians (both with and without genetic specialization), nurses, and genetic counselors. Various provider types may be combined, even within a single study, and bring different training (particularly around skills in providing psychological support and counseling and methods to impart educational components of genetic counseling) and time availability for services. Second, to date, measures of the effectiveness of genetic counseling outcomes have been broadly defined and have included knowledge, reproductive decisionmaking, behavior change, satisfaction, interpersonal measures, psychological support, aid in decisionmaking, and cost-effectiveness. Studies of outcomes also have been conflated by including the genetic testing process, and it is often difficult to ascertain whether an outcome is due to the genetic counseling service or the genetic testing process. Finally, genetic counseling services occur in various clinical settings (e.g., prenatal, pediatric, oncology, and other adult areas), making it even more difficult to select key outcomes to assess. At the end of this appendix is a comprehensive bibliography of references organized by topic, presenting key areas where effectiveness has been measured to date.

Knowledge has been one of the primary outcome measures for genetic counseling, and many studies have documented an increase in knowledge after genetic counseling and made comparisons to other educational modalities, including brochures, videos, and computer approaches.[112,113] Other studies have described the educational process by various providers.[114]

Obtaining relevant clinical data is yet another area where genetic counseling services may impact clinical care. Studies suggest that nongenetics professionals do not routinely take three-generation pedigrees. The incorporation of a pedigree taken by a genetic counselor or using measures designed and interpreted

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[109] ACMG. Folic acid and pregnancy. http://www.acmg.net/resources/policies/pol-011.asp [November 25, 2005].
[110] Massachusetts 2000 Session Laws. An Act relative to insurance and genetic testing and privacy protection. www.mass.gov/legis/laws/seslaw00/sl000254.htm [November 25, 2005].
[111] Michigan Public Health Code 333.17020. http://www.legislature.mi.gov/mileg.asp?page=getObject&objName=mcl-333-17020 [November 25, 2005].
[112] Bernhardt BA, Biesecker BB, Mastromarino CL. Goals, benefits, and outcomes of genetic counseling: Client and genetic counselor assessment. *Am J Med Genet* 2000. 94(3):189-97.
[113] Ciske et al. Genetic counseling and neonatal screening for cystic fibrosis: An assessment of the communication process. *Pediatrics* 2001. 107(4):699-705.
[114] For example, Bernhardt BA, et al. Prenatal genetic testing: Content of discussions between obstetric providers and pregnant women. *Obstet Gynecol* 1998. 91(5 Pt 1):648-55.

by a genetic professional pick up at least 20 percent of additional families at increased risk for genetic disorders.[115,116]

Key areas where genetic counseling services may impact clinical care are around informed consent and test interpretation, both of which are critical to informed patient decisionmaking. For example, Giardiello et al. found that only 16.9 percent of patients underwent written informed consent before adenosis polyposis coli (APC) genetic testing, and that 31.6 percent of APC test results were misinterpreted by nongenetics physicians.[117] Other studies suggest that when pregnant patients faced with a sex chromosome anomaly on amniocentesis receive information from genetics specialists v. nonspecialists (e.g., obstetricians or pediatricians), they receive more updated information and are less likely to undergo pregnancy termination.[118,119] Data also suggest that the higher the amount of negative information received, the more likely a prospective parent is to terminate an affected pregnancy.[120] As Braddock stated, there is a move toward increasing patient roles in decisionmaking, and health care systems need to find ways to increase it in clinical practice; genetic counseling services meet this increasing need.[121]

With regard to the cost-effectiveness of genetic counseling services, most studies bundle genetic counseling and testing, making it difficult to interpret study results.[122,123] Few studies assess a patient's willingness to pay for genetic counseling services, but Bernhardt et al. found that patients were willing to pay a mean of $200 to receive such services.[124] NSGC has contracted with researchers at the University of Washington to conduct a study that includes a literature review, development of a conceptual process-outcome cost model that illustrates the common points where expenditures by third-party payers could be incurred, and a Web-based survey of genetic counselors whose practices focus on prenatal care. Preliminary data from the empirical model suggest that the benefits to coverage of prenatal genetic counseling services come primarily from higher rates of risk identification, more informed deliveries, and lower costs.

The Importance of Reimbursing Genetic Counseling Services, Benefits in Doing So, and Potential Harms if Reimbursement Is Not Obtained. Once we have established the value of genetic counseling services and the qualifications of nonphysician providers to conduct such services, we can consider the benefits and barriers to obtaining coverage and reimbursement for such services. The issue of reimbursement for genetic

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[115] Frezzo TM, et al. The genetic family history as a risk assessment tool in internal medicine. *Genet Med* 2003. 5(2):84-91.
[116] Cohn GM, et al. The usefulness of a prenatal genetic questionnaire in genetic risk assessment. *Obstet Gynecol* 1996. 88(5): 806-10.
[117] Giardiello FM, et al. The use and interpretation of commercial APC gene testing for familial adenomatous polyposis. *N Engl J Med* 1997. 336(12):823-7.
[118] Abramsky L, et al. What parents are told after prenatal diagnosis of a sex chromosome abnormality: Interview and questionnaire study. *BMJ* 2001. 322(7284):463-6.
[119] Hall S, Abramsky L, and Marteau TM. Health professionals' reports of information given to parents following the prenatal diagnosis of sex chromosome anomalies and outcomes of pregnancies: A pilot study. *Prenat Diagn* 2003. 23(7):535-8.
[120] Ibid.
[121] Braddock CH et al. Informed decision making in outpatient practice: Time to get back to basics. *JAMA* 1999. 282(24):2313-20.
[122] Balmana J et al. Genetic counseling program in familial breast cancer: Analysis of its effectiveness, cost and cost-effectiveness ratio. *Int J Cancer* 2004. 112:647-52.
[123] Cohen D et al. Health economics and genetic service development: A familial cancer genetic example. *Familial Cancer* 2004. 3:61-7.
[124] Bernhardt BA, Biesecker BB, Mastromarino CL. Goals, benefits, and outcomes of genetic counseling: Client and genetic counselor assessment. *Am J Med Genet* 2000. 94(3):189-97.

counseling services is critical when we consider the impact on the genetics workforce. If genetic counseling services are not reimbursed, it likely will impact the access to services nationally, leading to a two-tiered system for genomic health care and increasingly unequal access to services.

Several key points are critical to understanding why it is so critical to obtain coverage and reimbursement for genetic counseling services at this juncture. Historically, many clinical genetic counseling services were provided by master's-level genetic counselors at centers that held maternal and child health block grants to fund the overarching provision of clinical services. With the funding base of these grants, the current imperative for billable services was minimized, since genetic counselor (and geneticist) salaries could be considered a component of grant funding. This funding support is decreasing, making it increasingly critical to establish other financial supports for genetic service provision. In 2004, 57 percent of genetic counselors reported billing for services in their supervising physician's name, 9 percent bill under their own name and the physician's name, and 14 percent reported not billing for services at all.[125] We are not aware of studies that have addressed the impact of these various billing practices. ISONG survey results show that 12 percent of genetic nurse specialists are nurse practitioners who can bill for services.

The 2004 NSGC professional status survey shows that 81 percent of genetic counselors currently work in a university medical center, public or private hospital, HMO, or private physician's office.[126] In the past 20 years, since data collection began, there has been a large relative decrease in genetic counselors reporting employment by university medical centers, and there is a slight decrease in overall health center employment.

According to data from NSGC, members see approximately 1.2 million clinical cases per year, and caseloads have been increasing approximately 5 percent per year since 2000. There has been a slight decrease in the percentage of genetic counselors reporting clinical care as a primary role (from 86 percent in 2002 to 83 percent in 2004), whereas research as a primary role is increasing (from 30 percent to 32 percent).[127] Many ISONG members also have moved away from providing patient care, as indicated by the percentage of time they spend in individual patient care. Sixty-two percent of ISONG members spend 20 percent or less of their time directly involved with patient care.[128] Results from the ISONG nursing survey show that 68 percent of genetic nurses spend some portion of their professional time each week participating in research.[129] Twenty-two percent were involved in research for 10 percent of their time or less, and 46 percent were involved for 10 percent or more.[130] Data from the survey indicated that genetic nurses are generally comfortable with the level of support they receive from their institutions. Fifty-three percent are satisfied with institutional assistance (financial, human resources, etc.), and 16 percent are very satisfied.[131] Regarding reimbursement, ISONG members deem the adequacy of reimbursement for genetic services to be poor/fair (69 percent and 70 percent, respectively).[132]

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[125] NSGC. 2004 Professional Status Survey. http://www.nsgc.org/careers/2004_PSS_Final_pw.pdf [November 25, 2005].
[126] Ibid.
[127] NSGC. Professional Status Surveys. http://www.nsgc.org/careers/pss_index.asp [November 25, 2005].
[128] Genetics Health Services Research Center, University of Maryland School of Medicine. Advanced Practice Nurses in Genetics: A Survey of ISONG Members.
[129] Ibid.
[130] Ibid.
[131] Ibid.
[132] Ibid.

Although the cause for this change in role diversification is not clear, it may be related to the increasing need to find salary support for those providing genetic counseling services, given the inability to generate significant billable service reimbursement. Additionally, if individuals providing genetic counseling services are required to spend increasing time on research or other administrative or teaching obligations to provide funding toward their salaries, the time available to provide clinical services will decrease, making it even more difficult to meet the growing need for clinical genetic counseling services.

Benefits Associated With Reimbursement for Nonphysician Genetic Counseling Services. As outlined by Avis Gibons, there are several reasons why insurers should consider reimbursement for genetic counseling services through nonphysician providers.[133] First, the use of genetic counseling services will lead to cost containment through the more appropriate use of genetic tests and appropriate test interpretation. For example, prenatal genetic counseling has been associated with reducing costs in high-risk or at-risk pregnancies.[134] Additionally, in the broader health dollar discussion, since genetic counselors typically bill at 50 to 80 percent of the level of physicians, cost containment will occur by using nonphysician providers for the time-intensive genetic counseling, education, and support process.[135]

An additional, not yet formally documented benefit of coverage for genetic counseling services includes the potential increase in number of service providers. Although this has not been broadly documented, Utah, the only State that currently licenses genetic counselors, saw a secondary benefit to licensure as it increased recognition of these nonphysician health care providers and allowed the genetic counselors to more easily be recognized as providers with third-party payers, satisfying compliance officers regarding "incident to" billing. In the 19 months after licensure was enacted, genetic counselors were recognized as providers by seven third-party payers and were allowed to bill under their attending physician by four additional insurers, with approximately 55 to 75 percent of charges being reimbursed.[136] Since 2002 Utah has observed an increase from 14 to 25 genetic counselors state-wide since licensure was enacted; much of this increase has been anecdotally attributed to the increasing recognition and ability to bill for clinical services.

One must also assess coverage and reimbursement discussions by considering the bundling of genetic counseling services with genetic testing. In the study by Gibons, 60.4 percent of respondents reported coverage for genetic counseling services, and 68.9 percent reported testing coverage, but 31.3 percent stated that genetic counseling services were provided only in conjunction with genetic testing, and only 8.9 percent required that genetic counseling accompany genetic testing.[137] This is somewhat in contrast to the recommendations by various States and insurers that suggest or require that genetic counseling services be provided in conjunction with testing.

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[133] Gibons A. Study results: Employer-based coverage of genetic counseling services. *Benefits Quarterly* 2004, pp. 48-68.
[134] Helitzer J. Genetic testing and prophylactic surgery: To slowly go where few have gone before. *Benefits Law J* 1999. 12:123-5.
[135] Gibons A. Study results: Employer-based coverage of genetic counseling services. *Benefits Quarterly* 2004, pp. 48-68.
[136] Cantrell M. Life after licensure: Our office's reimbursement experience. Presented at NSGC Annual Education Conference, October 2004.
[137] Gibons A. Study results: Employer-based coverage of genetic counseling services. Benefits Quarterly 2004, pp. 48-68.

Current Status. Some States and insurers are already proposing not only the inclusion of genetic counseling services in their recommended services but also reimbursement. Examples are:

- Washington State mandates Medicaid coverage for prenatal diagnosis genetic counseling and mandated benefits for prenatal genetic services.[138]
- Texas Medicaid developed billing codes and reimbursement levels for genetic evaluation and counseling services.[139] Example maximum fees range from $50.75 (for followup medical genetic counseling) to $152.25 for detailed health history and prenatal genetic counseling to $370.48 (for a detailed health history, comprehensive physical exam, and complex psychosocial assessment).
- Uniform Medical Plan in Washington State requires that genetic cancer susceptibility testing be accompanied by genetic counseling performed by a board-certified genetics professional.
- In 2002 Aetna developed protocols that cover not only genetic testing but also genetic counseling consultation by "qualified counselors and physicians."[140,141] Specifically, they state, "Aetna considers genetic counseling in connection with pregnancy management under plans with benefits for family planning medically necessary for evaluation of any of the following [list deleted]… Aetna considers appropriate genetic counseling unrelated to pregnancy in conjunction with covered genetic tests, and in accordance with the guidelines of the American College of Medical Genetics medically necessary."[142]

Several additional pieces of data may be useful in considering this issue

- The Washington State Department of Health developed a genetic services section plan for 2002-2005. One of the goals stated was to examine coverage of genetic services by the top 10 self-insured employers in the State.
- A survey of National Cancer Institute Comprehensive Cancer Centers in 1995 documented that half the Centers provided genetic services at that time, and that of those, 76.5 percent had a genetic counselor on staff, and 70.6 percent had a medical geneticist.[143] A survey assessing the changes over the past 10 years is currently being completed.[144]

Potential Harms if Reimbursement Is Not Available for Nonphysician Genetic Counseling Services. We are not aware of any studies or editorials that have addressed the potential harms that will occur if reimbursement and coverage do not become available for nonphysician genetic counseling providers. We are happy to discuss the anecdotal concerns, particularly around the impact on the workforce and subsequent access to genetic counseling services, at the Committee's request.

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[138] Washington State Department of Health. Genetic Services Section Plan. http://mchneighborhood.ichp.edu/wagenetics/WA-State-GeneticsStrategicPlan-9-30-02.pdf [November 25, 2005].
[139] 2005 Texas Medicaid Provider Procedures Manual. Section 21.3. http://www.tmhp.com/File%20Library/File%20Library/Provider%20Manuals/Texas%20Medicaid%20Provider%20Procedures/2005%20TMPPM-%20Individual%20Chapters/21_TMPPM05_Genetics.pdf [November 25, 2005].
[140] Aetna Press Release. Aetna recommends guidelines for access to genetic testing. http://www.aetna.com/news/2002/pr_20020617.htm [November 25, 2005].
[141] Rowe J. The financing of genetic technologies in the U.S. health care system. Presentation to SACGHS on June 11, 2003. http://www4.od.nih.gov/oba/SACGHS/meetings/June2003/Presentations/Rowe_t.pdf [November 25, 2005].
[142] Aetna. Clinical Policy Bulletin: Genetic Counseling. http://www.aetna.com/cpb/data/CPBA0189.html [November 25, 2005].
[143] Thompson JA et al. Genetic services for familial cancer patients: A survey of National Cancer Institute Cancer Centers. *JNCI* 1995. 87(19):1446-55.
[144] Judy Garber, Director, Cancer Risk and Prevention Program, Dana-Farber Cancer Institute. Personal communication, February 2005.

Summary and Recommendations

SACGHS is in the position to make recommendations to the Department of Health and Human Services regarding the future of genetic services in health care. With the extraordinary impact of genetic information on health and society, genetic service providers are in the position to provide information and health care services to the public. Currently, the structure exists to guide training programs to produce high-quality, certified genetics professionals, and licensure is being explored by a majority of States. However, without adequate reimbursement for genetic services, public health could be compromised by the provision of genetic services by uninformed health care providers without specialized training.

As our literature review has shown, genetic counseling has demonstrated value and is effective. Furthermore, providing coverage and reimbursement to nonphysician genetic counseling service providers will decrease costs and likely increase access. No studies currently exist on the potential harms if nonphysician genetic service providers are unable to obtain coverage and reimbursement for services. We strongly encourage SACGHS to make formal recommendations to:

- Recognize, through licensure and other mechanisms, nonphysician providers with expertise in genetics, as demonstrated by being credentialed by a national genetics organization appropriate for providers of genetic counseling services
- Advocate in all manners possible for the development of CPT codes that are specific to genetic counseling services for use by any qualified provider
- Support the funding of further studies to assess the value and effectiveness of genetic counseling services provided by nonphysicians

In conclusion, data are presented here that outline the qualifications, value, and effectiveness of genetic counselors, credentialed genetics nurses, and similarly trained health professionals. SACGHS can now provide recommendations at high levels that will assist with achieving reimbursement for nonphysician providers to allow high-quality, effective health care services. With reimbursement, these providers can become even more valuable in the financial realm of U.S. health care and allow more medical facilities to offer high-quality genetic services to the public.

Articles on the Value and Effectiveness of Genetic Counseling Services

Credential Programs

Cooksey JA. The genetic counselor workforce: Training programs, professional practice, and issues affecting supply and demand. Illinois Center for Health Workforce Studies, 2000.

Qualified Providers – Nurse and Genetic Counselors

Bernhardt BA et al. Evaluation of nurses and genetic counselors as providers of education about breast cancer susceptibility testing. *Oncol Nurs Forum* 2000. 27(1):33-9.

Qualified Providers – Genetic Nurse Credentialing

Association of Women's Health and Neonatal Nursing (2004). The role of the registered nurse as related to genetic testing. http://www.awhonn.org/awhonn/?pg=873-6230-6990-4730-5400-7430 [November 18, 2005].

Black RM, ed. Genetics Nursing Portfolios: A New Model for Credentialing. Washington, DC: American Nurses Association, 2004.

Calzone KA, Jenkins J, and Masny A. Core competencies in cancer genetics for advanced practice oncology nurses. *Oncol Nurs Forum* 2002, 29(9):1327-33.

Cook SS et al. Portfolio evaluation for professional competence: Credentialing in genetics for nurses. *J Prof Nurs* 2003. 19(2):85-90.

Greco K and Mahon SM. Genetics nursing practice enters a new era with credentialing. *Internet Journal of Advanced Nursing Practice* 2003. 5(2): http://www.ispub.com/ostia/index.php?xmlFilePath=journals/ijanp/vol5n2/genetics.xml [November 18, 2005].

International Society of Nurses in Genetics (ISONG) Genetics Nursing Credentialing Commission. What is a genetic nurse? 2003. http://64.78.6.122/resources/what_is.pdf [November 18, 2005].

ISONG. Statement on the scope and standards of genetics clinical nursing practice. Washington, DC: American Nurses Association, 1998.

ISONG. Informed decision-making and consent: The role of nursing. 2000, revised 2005. http://64.78.6.122/about/ps_consent.cfm [November 18, 2005].

ISONG. Genetic counseling for vulnerable populations: The role of nursing. 2002. http://64.78.6.122/about/ps_vulnerable.cfm [November 18, 2005].

Lea DH. Integrating genetics into baccalaureate and advanced nursing education. *Nurs Outlook* 2002. 50:167.

Monsen RB. Genetic nursing portfolios: A new model for credentialing. Washington, DC: American Nurses Association, 2004. http://nursingworld.org/books/pdescr.cfm?cnum=2#04GNP [November 18, 2005].

Oncology Nursing Society. The role of the oncology nurse in cancer genetic counseling. *Nurs Forum* 2000. 27(9):1348.

Oncology Nursing Society. Cancer predisposition genetic testing and risk assessment counseling. http://www.ons.org/publications/positions/CancerPredisposition.shtml [November 18, 2005].

Qualified Providers – Master's-Level Genetic Counselor Credentialing

American Society of Human Genetics Ad Hoc Committee on Genetic Counseling. Genetic counseling. *Am J Hum Genet* 1979. 27:240-2.

Fiddler MB et al. Practice-based competencies for accreditation of and training in graduate programs in genetic counseling. *J Genet Counsel* 1996. 5:113-21.

James et al. Professional preparation of individuals who provide genetic counseling services. *J Genet Couns* 1995. 4(1):49-63.

Kumars KW, et al. Genetics associates: Their training, role and function. A conference report. Washington, DC: U.S. Department of Health and Education and Welfare, 1979.

Marks JH and Richter ML. The genetic associate: A new health professional. *Am J Public Health* 1976. 66:388-90.

Smith A. Update on Master's genetic counseling training programs: Survey of curriculum content and graduate analysis summary. *J Genet Couns* 1993. 2(3):197-211.

Walker AP et al. Report of the 1989 Asilomar meeting on education in genetic counseling. *Am J Hum Genet* 1990. 46:1223-30.

Genetic Counselor Qualified Independent of Physician

Sommer A et al. Minimum guidelines for the delivery of prenatal genetics services. The evaluation of clinical services subcommittee, Great Lakes Regional Genetics Group. *Genet Med* 1999. 1(5):233-4.

Value and Effectiveness of Genetic Counseling

Abramsky L et al. What parents are told after prenatal diagnosis of a sex chromosome abnormality: Interview and questionnaire study. *BMJ* 2001. 322(7284):463-6.

Aktan-Collan K et al. Evaluation of a counselling protocol for predictive genetic testing for hereditary non-polyposis colorectal cancer. *J Med Genet* 2000. 37(2):108-13.

American Society of Clinical Oncology. Policy statement update: Genetic testing for cancer susceptibility. *J Clin Oncol* 2003. 21(12):2397-2406.

American Urological Association and American Society for Reproductive Medicine. Infertility: Report on Optimal Evaluation of the Infertile Male. 2001. http://www.auanet.org/timssnet/products/guidelines/main_reports/optimalevaluation.pdf [November 18, 2005].

Balmana J et al. Genetic counseling program in familial breast cancer: Analysis of its effectiveness, cost and cost-effectiveness ratio. *Int J Cancer* 2004. 112:647-52.

Bernhardt BA, Biesecker BB, and Mastromarino CL. Goals, benefits, and outcomes of genetic counseling: Client and genetic counselor assessment. *Am J Med Genet* 2000. 94(3):189-97.

Ciske DJ et al. Genetic counseling and neonatal screening for cystic fibrosis: An assessment of the communication process. *Pediatrics* 2001. 107(4):699-705.

Cohn GM et al. The importance of genetic counseling before amniocentesis. *J Perinatol* 1996. 16(5):352-9.

Cohn GM et al. The usefulness of a prenatal genetic questionnaire in genetic risk assessment. *Obstet Gynecol* 1996. 88(5):806-10.

Cunningham GC and Tompkinison DG. Cost and effectiveness of the California triple marker prenatal screening program. *Genet Med* 1999. 1(5):199-206.

Fanos JH, Davis J, and Puck JM. Sib understanding of genetics and attitudes toward carrier testing for X-linked severe combined immunodeficiency. *Am J Med Genet* 2001. 98(1):46-56.

Fanos JH and Gatti RA. A mark on the arm: Myths of carrier status in sibs of individuals with ataxia-telangiectasia. *Am J Med Genet* 1999. 86(4):338-46.

Fanos JH and Johnson JP. Perception of carrier status by cystic fibrosis siblings. *Am J Hum Genet* 1995. 57(2):431-8.

Fanos JH. Developmental tasks of childhood and adolescence: Implications for genetic testing. *Am J Med Genet* 1997. 71(1):22-8.

Farrell M, Certain L, and Farrell P. Genetic counseling and risk communication services of newborn screening programs. *Arch Pediatr Adolesc Med* 2001. 155(2):120-6.

Frezzo TM et al. The genetic family history as a risk assessment tool in internal medicine. *Genet Med* 2003. 5(2):84-91.

Hall S, Abramsky L, and Marteau TM. Health professionals' reports of information given to parents following the prenatal diagnosis of sex chromosome anomalies and outcomes of pregnancies: A pilot study. *Prenat Diagn* 2003. 23(7):535-8.

International Huntington Association (IHA) and the World Federation of Neurology (WFN) Research Group on Huntington's Chorea. Guidelines for the molecular genetics predictive test in Huntington's disease. *Neurology* 1994. 44(8):1533-6.

Koscica KL et al. Assessing genetic risk: Comparison between the referring obstetrician and genetic counselor. *Am J Obstet Gynecol* 2001. 185:1033-4.

Loescher L. Nursing roles in cancer prevention position statements. *Semin Oncol Nurs* 2004. 20(2):111-20.

Macleod R et al. Patient's perceptions of what makes genetic counseling effective: An interpretive phenomenological analysis. *J Health Psychol* 2002. 7(2):145-156.

Mahon SM and Greco K. Nurses who provide genetics counseling need ongoing education and certification. *Oncol Nurs Forum* 2003. 30(3):361-2.

Morris KT et al. Genetic counseling impacts decision for prophylactic surgery for patients perceived to be at high risk for breast cancer. *Am J Surg* 2001. 181(5):431-3.

Pelias MZ and Shaw MW. Medicolegal aspects of prenatal diagnosis. In: Milunsky A, ed. Genetic Disorders and the Fetus Diagnosis, Prevention and Treatment, 3rd ed. Baltimore: Johns Hopkins University Press, 1992.

Robinson A and Linden MG. Clinical Genetic Handbook. Boston: Blackwell Scientific Publications, 1993.

Rowley PT et al. Prenatal genetic counseling for hemoglobinopathy carriers: A comparison of primary providers of prenatal care and professional genetic counselors. *Am J Hum Genet* 1995. 56(3):769-76.

Saleem R et al. Variables influencing parental perception of inherited metabolic diseases before and after genetic counselling. *J Inherit Metab Dis* 1998. 21(7):769-80.

Scheuer L et al. Outcome of preventive surgery and screening for breast and ovarian cancer in BRCA mutation carriers. *J Clin Oncol* 2002. 20(5):1260-8.

Skirton H et al. Genetic counsellors: A registration system to assure competence in practice in the United Kingdom. *Community Genet* 2003. 6(3):182-3.

Smyth A. Value of genetic counseling: A parent's view. *BMJ* 2001. 322:1071.

Todora H et al. Perceptions of genetic risk assessment and education among first-degree relatives with colorectal cancer patients and implications for physicians. *Family Pract* 2001. 18(4):367-72.

Tyler A and Harper P. Attitudes of subjects at risk and their relatives towards genetic counselling in Huntington's chorea. *J of Med Genet* 1983. 20:179-88.

Visovsky C. Genetics counseling. A basic competency for nurses? *Nurs Leadersh Forum* 1999. 4(2):40-43.

Vogel WH. The advanced practice nursing role in a high-risk breast cancer clinic. *Oncol Nurs Forum* 2003. 30(1):115-22.

Williams JK et al. Genetic counseling outcomes validation by genetics nurses in the UK and US. *J Nurs Scholarsh* 2001. 33(4):369-74.

Health Care Provider Knowledge Problems – Need for Trained Professionals

Doksum T et al. Does knowledge about the genetics of breast cancer differ between nongeneticist physicians who do or do not discuss or order BRCA testing? *Genet Med* 2003. 5(2):99-105.

Fanos JH and Mackintosh MA. Never again joy without sorrow: The effect on parents of a child with ataxia-telangiectasia. *Am J Med Genet* 1999. 87(5):413-9.

Firth HV and Lindenbaum RH. UK clinicians' knowledge of and attitudes to the prenatal diagnosis of single gene disorders. *J Med Genet* 1992. 29(1):20-3.

Geller G and Holtzman NA. Implications of the human genome initiative for the primary care physician. *Bioethics* 1991. 5(4):318-25.

Geller G et al. Incorporation of genetics in primary care practice: Will physicians do the counseling and will they be directive? *Arch Fam Med* 1993. 2(11):1119-25.

Geller G et al. Physicians' attitudes toward disclosure of genetic information to third parties. *J Law Med Ethics* 1993. 21(2):238-40.

Giardiello FM et al. The use and interpretation of commercial APC gene testing for familial adenomatous polyposis. *N Engl J Med* 1997. 336(12):823-7.

Greendale K and Pyeritz RE. Empowering primary care health professionals in medical genetics: How soon? How fast? How far? *Am J Med Genet* 2001. 106(3):223-32.

Hayflick SJ et al. Primary care physicians' utilization and perceptions of genetics services. *Genet Med* 1998. 1(1):13-21.

Hayflick SJ et al. Role of primary care providers in the delivery of genetics services. *Community Gene* 1998. 1:18-22.

Hofman KJ et al. Physicians' knowledge of genetics and genetic tests. *Acad Med* 1993. 68(8):625-32.

Holtzman NA. The interpretation of laboratory results: The paradoxical effect of medical training. *J Clin Ethics* 1991. 2(4):241-3.

Holtzman NA. Primary care physicians as providers of frontline genetic services. *Fetal Diagn Ther* 1993. 8(suppl 1):213-9.

Jancin B. Failure to offer CF screen risks liability. *Ob Gyn News* 1998, p.29.

Kumar S and Gantley M. Tensions between policy makers and general practitioners in implementing new genetics: Grounded theory interview study. *BMJ* 1999. 319(7222):1410-3.

Lapham EV et al. The gap between practice and genetics education of health professionals: HuGEM survey results. *Genet Med* 2000. 2(4):226-31.

Lynch HT et al. Failure to diagnose hereditary colorectal cancer and its medicolegal implications: A hereditary nonpolyposis colorectal cancer case. *Dis Colon Rectum* 1999. 42(1):31-5.

Naylor EW. Genetic screening and genetic counseling: Knowledge, attitudes, and practices in two groups of family planning professionals. *Soc Biol* 1995. 22(4):304-14.

Ramsey PG et al. History-taking and preventive medicine skills among primary care physicians: An assessment using standardized patients. *Am J Med* 1998. 104(2):152-8.

Rose PW et al. Referral of patients with a family history of breast/ovarian cancer--GP's knowledge and expectations. *Fam Pract* 2001. 18(5):487-90.

Rowley PT and Loader S. Attitudes of obstetrician-gynecologists toward DNA testing for a genetic susceptibility to breast cancer. *Obstet Gynecol* 1996. 88(4 Pt 1):611-5.

Rowley PT et al. Cystic fibrosis carrier screening: Knowledge and attitudes of prenatal care providers. *Am J Prev Med* 1993. 9(5):261-6.

Ruo L et al. Limitations of family of cancer history assessment at initial surgical consultation. *Dis Colon Rectum* 2001. 44(1):98-104.

Sadler M. Serum screening for Down's syndrome: How much do health professionals know? *Br J Obstet Gynaecol* 1997. 104(2):176-9.

Shalev O et al. Inadequate utilization of routine electronic RBC counts to identify beta thalassemia carriers. *Am J Public Health* 1998. 78(11):1476-7.

Stratakis CA et al. Molecular genetics in pediatric training: How much do we really know? *Md Med J* 1995. 44(3): 210-213.

Strom CM et al. Cystic fibrosis screening: Lessons learned from the first 320,000 patients. *Genet Med* 2004. 6(3):136-40.

Summerton N and Garrood PV. The family history in family practice: A questionnaire study. *Fam Pract* 1997. 14(4):285-8.

Sweet KM, Bradley TL, and Westman JA. Identification and referral of families at high risk for cancer susceptibility. *J Clin Oncol* 2002. 20(2):528-37.

Taylor MRG. A survey of chairpersons of departments of medicine about the current and future roles of clinical genetics in internal medicine. *Genet Med* 2003. 5(4):328-31.

Vastag B. Cystic fibrosis gene testing a challenge. *JAMA* 2003. 289(22):2923-4.

Wilkins-Haug L. Obstetrician-gynecologists' opinions and attitudes on the role of genetics in women's health. *J Women's Health & Gender-Based Med* 2000. 9(8):873-9.

Reimbursement for Genetic Counseling Services

Cohen D et al. Health economics and genetic service development: A familial cancer genetic example. *Familial Cancer* 2004. 3:61-7.

Gibons A. Study results: Employer-based coverage of genetic counseling services. *Benefits Quarterly* 2004, p.48-68.

Helitzer J. Genetic testing and prophylactic surgery: To slowly go where few have gone before. *Benefits Law J* 1999. 12:123-5.

2005 Texas Medicaid Provider Procedures Manual. Section 21.3. http://www.tmhp.com/File%20Library/File%20Library/Provider%20Manuals/Texas%20Medicaid%20Provider%20Procedures/2005%20TMPPM-%20Individual%20Chapters/21_TMPPM05_Genetics.pdf [November 18, 2005].

Licensure Reasonable and Needed

Berwick DM. A user's manual for the IOM's Quality Chasm Report. *Health Affairs* 2002. 21(3):80-90.

Vance A. Licensing genetic counselors holds promise for higher quality, more cost-effective service for patients. *Gene Letter* 2000. 1(10).

Harms Due to Lack of Billing for Genetic Counseling

No literature found

Other

Association of Professors of Human or Medical Genetics. Clinical objectives in medical genetics for undergraduate medical students. *Genet Med* 1998. 1:54-5.

Bernhardt BA et al. Prenatal genetic testing: Content of discussions between obstetric providers and pregnant women. *Obstet Gynecol* 1998. 91 (5 Pt 1):648-55.

Berry RJ et al. Birth weight-specific infant mortality due to congenital abnormalities, 1960 and 1980. *Public Health Report* 1987. 102:171-81.

Blumenthal D et al. The duration of ambulatory visits to physicians. *J Fam Pract* 1999. 48(4):264-71.

Bonadona V et al. Cancer patients who experienced diagnostic genetic testing for cancer susceptibility: Reactions and behavior after the disclosure of a positive test result. *Cancer Epidemiol Biomarkers Prev* 2002. 11(1):97-104.

Braddock CH et al. Informed decision making in outpatient practice: Time to get back to basics. *JAMA* 1999. 282(24): 2313-20.

Collins FS. Shattuck Lecture: Medical and societal consequences of the Human Genome Project. *New Engl J Med* 1999. 341(1):28-37.

Collins F and Guttmacher A. Genetics moves into the medical mainstream. *JAMA* 2001. 286(18):2322-4.

Collins FS and McKusick VM. Implications of the Human Genome Project for medical science. *JAMA* 2001. 285(5):540-4.

Cornel MC et al. Comparison of couples referred and not referred for genetic counseling in a genetic clinic after the birth of a child with a congenital anomaly: A study in a population in the northeastern Netherlands. *Am J Med Genet* 1992. 42(3): 387-92.

Deftos LJ. The evolving duty to disclose the presence of genetic diseases to relatives. *Acad Med* 1998. 73:962-8.

DeMichele A and Weber B. Risk management in BRCA1 and BRCA2 mutation carriers: Lessons learned, challenges posed. *J Clin Oncol* 2002. 20(5):1164-6.

Dickson D. British insurers continue with genetic tests. *Nat Med* 1999. 5(9):974.

Emery AEH and Rimoin DL. Principles and Practice of Medical Genetics, 2nd ed. New York: Churchill Livingstone, 1990.

Epstein CJ et al. Genetic counseling. *Am J Hum Genet* 1975. 27(2):240-2.

Faden RR et al. Attitudes of physicians and genetics professionals toward cystic fibrosis carrier screening. *Am J Med Genet* 1994. 50(1):1-11.

Fanos JH and Johnson JP. Barriers to carrier testing for adult cystic fibrosis sibs: The importance of not knowing. *Am J Med Genet* 1995. 59(1):85-91.

Geller G et al. Decision-making about breast cancer susceptibility testing: How similar are the attitudes of physicians, nurse practitioners, and at-risk women? *J Clin Oncol* 1998. 16(8):2868-76.

Godard B et al. Strategies for consulting with the community: The cases of four large-scale genetic databases. *Sci Eng Ethics* 2004. 10(3):457-77.

Gollust S et al. Limitations of direct-to-consumer advertising for clinical genetic testing. *JAMA* 2002. 288(14):1762-7.

Grembowski DE et al. Managed care and physician referral. *Med Care Res Rev* 1998. 55(1):3-31.

Halliday J et al. Use of record linkage between a statewide genetics service and a Birth Defects/Congenital Malformations Register to determine use of genetic counselling services. *Am J Med Genet* 1997. 72(1):3-10.

Hamann HA et al. Attitudes toward the genetic testing of children among adults in a Utah-based kindred tested for a BRCA1 mutation. *Am J Med Genet* 2000. 92(1):25-32.

Holtzman NA. Is public health ready for genetics? *Arch Pediatr Adolesc Med* 2001. 155(2):117-8.

Kahn J. Is there a cure for cancer ads? A TV campaign for breast cancer gene tests has doctors alarmed. *Fortune* February 19, 2003.

Kessler S et al. Psychological aspects of genetic counseling. III. Management of guilt and shame. *Amer J Med Genet* 1984. 17:673-97.

Kussman J et al. Current and desired roles in the provision of genetic services among family physicians in the United States. *J Genet Couns* 2004. 13(6):543-4

Leff M. The best of health: 275 questions you've always wanted to ask your doctor. *Consumers Union* 1998.

Lerman C et al. BRCA1 testing in families with hereditary breast-ovarian cancer: A prospective study of patient decision making and outcomes. *JAMA* 1996. 275(24):1885-92.

Lilford RJ et al. Effect of using protocols on medical care: Randomised trial of three methods of taking an antenatal history. *BMJ* 1992. 305(6863):1181-4.

MacLeod et al. Patients' perceptions of what makes genetic counselling effective: An interpretative phenomenological analysis. *J Health Psychol* 2002. 7:145-56.

Marcus A. First ad campaign touts genetic screening for cancer, but critics say testing without counseling first can be harmful. Health on the Net Foundation, October 15, 2002.

McConkie-Rosell A et al. Carrier testing in the fragile X syndrome: Attitudes and opinions of obligate carriers. *Am J Med Genet* 1997. 68(1):62-9.

McGovern MM, Benach M, and Zinberg R. Interaction of genetic counselors with molecular genetic testing laboratories: Implications for non-geneticist health care providers. *Am J Med Genet* 2003. 119A:297-301.

McKusick VA. Mendelian Inheritance in Man: A Catalog of Human Genetics and Genetic Disorders, 11th ed. Baltimore: Johns Hopkins University Press, 1994.

Oppenheim D et al. The psychological burden inflicted by multiple cancer in Li-Fraumeni families: Five case studies. *J Genet Couns* 2001. 10(2):169-83.

Pyeritz RE. Economic considerations in providing clinical genetic services. Birth Defects Original Article Series 1999. 26(2):67-73.

Quaid K. Presymptomatic testing for Huntington disease: Recommendations for counseling. *J Genet Couns* 1992. 1(4):277-302.

Resta R. Eugenics and nondirectiveness in genetic counseling. *J Genet Couns* 1997. 6(2):255-8.

Ribeiro M et al. Incorrect genetic counseling of a couple with beta-thalassemia, due to incomplete testing. *Am J Hum Genet* 1993. 52:842-3.

Scheuner MT et al. Family history: A comprehensive genetic risk assessment method for the chronic conditions of adulthood. *Am J Med Genet* 1997. 71:315-24.

Scheuner MT. Family history: Where to go from here. *Genet Med* 2003. 5(2):66-9.

Schoonmaker M, Bernhardt B, and Holtzman NA. Coverage of new genetic technologies: What matters to insurers? *Am J Hum Genet* 1996. 59(4):A3.

Scriver CR et al. The frequency of genetic disease and congenital malformation among patients in a pediatric hospital. *Canadian Med Assoc J* 1993. 108:1111-5.

Silverman PH. Commerce and genetic diagnostics. Hastings Center Report. 25(3 suppl):S15-7.

St Peter RF et al. Changes in the scope of care provided by primary care physicians. *N Engl J Med* 1999. 341(26):1980-5.

St Peter RF et al. The scope of care expected of primary care physicians: Is it greater than it should be? *Issue Brief Cent Stud Health Syst Change* 1999. (24):1-4.

Strom CM et al. Cystic fibrosis screening: Lessons learned from the first 320,000 patients. *Genet Med* 2004. 6(3):136-40.

Strom CM et al. Cystic fibrosis screening using the College panel: Platform comparison and lessons learned from the first 20,000 samples. *Genet Med* 2002. 4(4):289-296.

Thompson JA et al. Genetic services for familial cancer patients: A survey of National Cancer Institute Cancer Centers. *JNCI* 1995. 87(19):1446-55.

Walpole IR et al. Evaluation of a project to enhance knowledge of hereditary diseases and management. *J Med Genet* 1997. 34(10):831-7.

Weitz R. Barriers to acceptance of genetic counseling among primary care physicians. *Soc Biol* 1979. 26(3):189-97.

Wood T. Risk assessment crucial for cancer patients. 2001 American Society of Human Genetics Meeting, October 15, 2001.

Yoong AF et al. Audit of compliance with antenatal protocols. *BMJ* 1992. 305(6863):1184-6.

Appendix C

Public Comments

The following individuals and organizations responded to an April 2005 request for public comment on an earlier version of this report.

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American Academy of Actuaries

American Association for Clinical Chemistry

American Board of Genetic Counseling

American Clinical Laboratory Association

American College of Medical Genetics

American Psychoanalytic Association

American Society for Microbiology

American Society of Human Genetics

America's Health Insurance Plans

Association for Molecular Pathology

BlueCross BlueShield Association

Boston Healthcare Associates

College of American Pathologists

Genetic Alliance

Genetic Task Force of Illinois

GroupHealth Permanente

Institute for Health Freedom

International Society of Nurses in Genetics

National Society of Genetic Counselors

Oncology Nursing Society

Personalized Medicine Coalition