



GENETIC SERVICES POLICY PROJECT^{*}

YEAR ONE REVIEW

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INTRODUCTION AND BACKGROUND

The Genetic Services Policy Project is a collaborative effort involving the Health Resources and Services Administration Maternal and Child Health Bureau, the Washington State Department of Health Genetic Services Section, the University of Washington Resource Center for Health Policy, and the Fred Hutchinson Cancer Research Center. The project began in June 2004 and is supported by grant numbers U35MC02601 and U35MC02602.

GSPP combines two complementary proposals titled Delivering Genetic Services and Translational Genetics Public Policy. The combined goals of the projects are to:

1. Characterize and evaluate the current model for genetic services delivery, focusing on the economic, legal, cultural, and policy aspects;
2. Characterize and evaluate potential alternative models for delivery of genetic services; and
3. Identify and assess options for public policies that promote cost effective, accessible, and equitable delivery of genetic services.
4. Develop an agenda to translate genetic research and information into practice;
5. Discuss and develop consensus about the most effective methods to translate genetic services information and research; and
6. Disseminate genetic services information and research findings to a wide range of decision makers involved in the financing, delivery, and consumption of genetic services.

These goals will be achieved through the completion of an exhaustive literature review, collection of qualitative data from key informant interviews, execution of legal, economic, and policy analyses and case studies, and interaction with key delivery system stakeholders.

GSPP project investigators and staff have diverse backgrounds and experiences and share a commitment to the successful delivery of genetic services. Project investigators and personnel are listed in Table 1. Over 25 representatives from key stakeholder groups serve as advisors and consultants to GSPP. The advisors and consultants are listed in Table 2.

YEAR ONE PROJECTS AND ACTIVITIES

Project personnel and investigators pursued several activities during year one of the project that were consistent with the stated goals and objectives. These included multiple literature reviews, profiles of state genetic service programs, gathering stakeholder perspectives, building collaborations with other projects, developing a Web site, monitoring emerging government or trade organizations' reports, and planning and executing two advisory committee meetings.

Literature Reviews

Expenditures: One of the literature reviews conducted by project personnel describes the current pool of information regarding expenditures for genetics services, which include genetic testing, diagnosis of genetic conditions, and treatments for individuals with genetic disorders. This review also identifies the limitations of the sources of information that are available and

concludes with possible sources that would enrich future discussions and research on genetic service utilization and expenditures. The paper is being prepared for publication and will be made available after publication.

Cost Effectiveness Analyses: Project personnel also reviewed the literature for articles describing original research on the cost effectiveness of genetic services. This review shows that there are relatively few economic evaluations for genetic services, and most are clustered in specific disease areas. The overall quality of the analyses was high, but varied widely. The authors concluded that most of the shortcomings of existing analyses are easy to address. For example, the relevance of these studies can be improved by using recommended measures of outcome. See Appendix A for an abstract of this review. We anticipate that the complete review will be published in *Genetics and Medicine* in late 2005 or early 2006. In addition, the abstract was accepted for presentation at two meetings: the International Health Economics Association, Barcelona, Spain, July 2005 and the International Society for Pharmacoeconomics and Outcomes Research, Washington DC, May 2005.

Billing and Reimbursement for Genetic Counselors: Project personnel contributed to a literature review funded by the National Society of Genetic Counselors (NSGC) on billing and reimbursement for genetic counselors. The purpose of this review was to develop a foundation for understanding the role of genetic counselors in the prenatal setting, how their services are accessed by patients, and how they are reimbursed for these services. The authors also looked for evidence about how the delivery of genetic services differs between genetic counselors and other providers of similar services. The review was also intended to inform recommendations regarding third party coverage for the genetic counseling profession. Appendix B is an annotated bibliography from this review.

Comprehensive Literature Database: During year one, project personnel began to develop a comprehensive literature database. The database will include published articles and reports from the scientific, health services, economic, legal, and social science literature. Bibliographies will be posted on the GSPP Web site during year two of the project.

Profiles of State Genetic Services

During year one, project personnel began to compile information on publicly funded and or administered genetic services programs. Eight states, one from each of the regional newborn screening collaboratives plus Washington State, were included in a pilot project to assess the feasibility of collecting specific information and the acceptability of different formats for presenting the information. For each state, we sought information about funding sources, organizational structure, provider capacity, political and legal environments, newborn screening programs, and populations served.

The pilot project resulted in four products: a white paper describing the methodology and results of our research, a compilation of data on each state, and two slide presentations. Copies of each of these items can be viewed online with the materials from the April 2005 Advisory Committee Meeting (<http://depts.washington.edu/genpol/advisory/>). This pilot project involving eight states suggests that state genetic services delivery systems vary in organizational structure and rely on a

wide variety of funding sources. We also found that sources of comprehensive data about genetic service utilization and expenditures are scarce. During year two, project personnel will collect information on additional states and complete an analysis of how state genetic services programs are structured and funded. Ultimately, we hope to identify efficient delivery models and make recommendations for improving the delivery of publicly funded and administered genetics programs.

Stakeholder Perspectives

An important component of GSPP is the stakeholder advisory committee. During year one, project personnel sought to gain knowledge about the perspectives of each of the key stakeholders represented on the advisory committee. The discussion at the April 2005 Advisory Committee Meeting proved to be an opportunity for the advisors to share their perspectives on issues related to coverage and reimbursement for genetic services, provider capacity and training, and the population that receives genetic services. The meeting summary in Appendix E provides a synopsis of this discussion. Elements from this discussion will be combined with results from a literature review and individual interviews with key informants to provide a comprehensive review of stakeholders' common and conflicting interests.

Building Collaborations with Other Projects

Project personnel worked very closely with personnel from other projects during year one. For example, some of the project investigators for GSPP also work for the Center for Excellence in ELSI Research (CEER) at the University of Washington. CEER and GSPP collaborate on shared issues and communicate about independent projects that may be of mutual interest. Work done by CEER personnel on newborn screening programs helped inform GSPP's profiles of state genetic services work. In addition, CEER and GSPP are collaborating to produce several case studies that will be completed in year two.

Developing a Web Site

The Internet is an important tool for disseminating information. During year one, project personnel worked to develop a Web site (<http://depts.washington.edu/genpol/>). The Web site serves as a means to make materials and information available to the public as well as to project personnel and advisors.

Commissioned Reports

Government agencies, professional organizations, and not for profit advocacy groups occasionally commission exploratory reports on issues related to genetic services. In March 2005, the Secretary's Advisory Committee on Genetics, Health and Society released a draft of one such report titled, "Coverage and Reimbursement for Genetic Tests and Services." GSPP personnel reviewed the report, drafted a summary of it for the project's advisors, and wrote a letter to the committee in response to a request for public comments. The letter commended the Committee for its work and encouraged it to seek diverse perspectives on the issues addressed in the report. A copy of the summary and letter are included in Appendix C and the full report can

be viewed online at <http://depts.washington.edu/genpol/docs/SACGHS-ReportDraftApril2005.pdf>.

Planning and Executing Two Advisory Committee Meetings

Members of the GSPP advisory committee are located throughout the country, and in order to encourage their participation in the project and encourage a dynamic exchange of ideas and information, GSPP personnel organize at least one advisory committee meeting per year. In year one, we held two advisory committee meetings.

The first meeting was held in September 2004. This meeting provided an opportunity for the project's personnel to introduce the project's goals and objectives and describe how we planned to achieve them. It was also an opportunity to create small discussion groups and receive input from the advisors about how to structure the project and what types of activities to pursue. The advisors also received presentations from Lee Newcomer from Vivius about consumer choice in health care and Brett Davis from IBM about the development of information technology and its role in health care delivery. An agenda and a meeting summary are included in Appendix D, and a slide presentation given at the meeting can be viewed online at <http://depts.washington.edu/genpol/advisory/>.

The second meeting was held in April 2005. The purpose of this meeting was to share the results from the profiles of state genetic programs pilot project and seek feedback from the advisors about usefulness of the information being collected. In addition, the meeting included two presentations: Don Kemper from Healthwise gave a presentation on information therapy and its role in health care, and Frank Gilliam talked about framing issues and communicating complicated concepts. Time was allotted for a discussion of emerging themes such as who provides genetic services, how providers are paid, how providers are trained, and who receives genetic services. An agenda and a meeting summary are included in Appendix E.

APPENDIX A

Title: A systematic review of economic evaluations of genetic testing technologies

Authors: Scott Ramsey (sramsey@fhcrc.org), Josh Carlson (jcarlson@aol.com), Nora Henrikson (nhenriks@fhcrc.org), David Veenstra (veenstra@u.washington.edu)

Background: Genetic test technologies offer hope for early diagnosis and identification of persons at risk for serious diseases. Stemming in large part from the sequencing of the human genome, numerous genetic tests have been introduced into clinical practice. Because many of these tests are costly and applicable to large populations (e.g., screening all newborns for cystic fibrosis), evaluations of the cost-effectiveness of these technologies is important.

Objectives: To conduct a systematic search for and review of economic evaluations of genetic testing technologies.

Methodology: Literature searches were performed using PubMed, Proquest, LexisNexis, Expanded Academic Index, The Harvard Review of Economic Analyses, PsycINFO, National Institute for Clinical Excellence, and The Canadian Council on Technology Assessment in Health Care. For resources amenable to mesh searching, the mesh terms included were: economic(s) and/or cost(s), combined with genetic, gene, and/or genotype. Searches were limited to English language with publication dates from 1990 to present. Selection criteria included original articles in cost-effectiveness (as defined by Drummond et al, Oxford Press 1997) in genetic services. To be considered as a genetic service, either the disease or condition had to be primarily genetic or involve a genetic test, defined as the analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes or karyotypes for clinical purposes. Articles were categorized by clinical category and type of economic study (e.g., cost-utility, cost-benefit), and graded independently by the authors using CEA study quality system developed by Chiou et al (Med Care, 2003;41:32).

Results: A total of 149 abstracts were retrieved using the search terms; 63 met selection criteria. Types of economic studies were as follows, cost-utility studies (25%); cost-benefit (19%); cost-minimization (6%); cost-effectiveness analyses (59%). Clinical categories were as follows: preconception carrier testing (8%); prenatal diagnosis (40%); adult testing (57%). (Totals >100% due to some studies having more than one type of analysis). The studies considered interventions for 26 different medical conditions. Study quality using the Chiou et al grading method ranged from 43-100 (average 82). Cost-utility studies were of highest quality (mean 91); cost-minimization studies were of lowest quality (mean 63). Adult studies had the highest rating (mean 86); preconception testing studies were lowest in quality (mean 74). Intraclass correlation among raters was 0.82 (CI 0.70-0.89).

Conclusions: A substantial number of economic analyses have been published in human genetics across a wide range of conditions. A large proportion of studies do not include a measure of benefit that facilitates comparison with other medical interventions. Study quality varied widely. Priority areas for the field include increasing quality and uniformity of measures of outcome.

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APPENDIX B

Annotated Bibliography from NSGC Sponsored Literature Review on Billing and Reimbursement for Genetic Counselors

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Aalfs CM, et al. (2003).	Referral for genetic counselling during pregnancy: limited alertness and awareness about genetic risk factors among GPs.	<i>Family Practice</i> 20: 135-41.	2000-2001	The of 100 Dutch GPs women who received genetic counseling	1	Genetic counseling
Abramsky L, et al., (2001).	What parents are told after prenatal diagnosis of a sex chromosome abnormality: interview and questionnaire study.	<i>BMJ</i> 322: 463-6.	1998-1999	23 couples informed that a sex chromosome anomaly was identified in an otherwise anatomically normal and viable fetus; 29 health professionals who had previously made such a diagnosis.	3	Counseling; prenatal diagnosis
Ad Hoc Committee on genetic Testing/Insurance Issues (1995).	Genetic Testing and insurance.	<i>Am J Hum Genet.</i> 56(1):327-31.	Committee opinion.		A	Geneti3 testing 1nd insur1n3e
American College of Obstetricians and Gynecologists (1997).	Routine Ultrasound in Low-Risk Pregnancy.	<i>Practice Patterns</i> No. 5.	Review of evidence-based literature relating to the use of ultrasound in routine prenatal care.		A	Re3eived from 13OG Resour3e 3enter
American College of Obstetricians and Gynecologists (2001).	Prenatal Diagnosis of Fetal Chromosomal Abnormalities.	<i>Practice Bulletin</i> , No. 27.	Review of ACOG standards of care for prenatal screening and diagnosis.		A	Re3eived from 13OG Resour3e 3enter
American Society for Human Genetics Ad Hoc Committee on Genetic Counseling. (1975).	Genetic Counseling.	<i>Am J Hum Genet</i> 27: 240-242.	Committee position.		A	Geneti3 3ounseling
Balinsky W and Zhu C (2004)	Pediatric Cystic Fibrosis: Evaluating Costs and Genetic Testing	<i>Journal of Pediatric Health Care</i>	Review of literature and data on pediatric cystic fibrosis.		B	Geneti3 testing 3osts

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Benn, PA, et al., (2004).	Changes in the Utilization of Prenatal Diagnosis	<i>Obstet Gynecol</i> 103(6): 1255-60.	1991-2002	All amniotic fluid and CVS samples processed by the University of Connecticut Health Center.	1	Prenatal diagnosis; patient utilization
Bernhardt B and Pyeritz R. (1989).	The economics of clinical genetics services. III Cognitive genetics services are not self-supporting	<i>Am J Hum Genet</i> 44: 288-293.	Unknown	Four clinical genetics centers in the U.S.	1	Genetic services; economics
Bernhardt B, Biesecker B, Mastromarino C. (2000).	Goals, Benefits, and Outcomes of Genetic Counseling: Client and Genetic Counselor Assessment	<i>Am J Med Genet</i> 94: 189-197	Unknown	16 genetic counselors and 19 genetic counseling clients	1	Genetic counseling outcomes
Bernhardt BA, et al. (1992)	The economics of clinical genetic services. IV Financial Impact of Outpatient genetic services on an Academic Institution	<i>Am J Hum Genet</i> 50: 84-91.	1983	Charges and payments for 100 patients at five academic genetic clinics	1	Genetic Services
Bernhardt BA, et al., (1998).	Prenatal Genetic Testing: Content of Discussions Between Obstetric Providers and Pregnant Women.	<i>Obstet Gynecol</i> 91: 648-55.	Not reported	The first prenatal visit of 169 women with 21 obstetricians and 19 CNMs	1	Prenatal genetic testing; content
Biesecker BB (2001).	Goals of genetic counseling.	<i>Clinical Genetics</i> . 60: 323-30.	Review		A	Geneti3 3ounseling
Bourguignon A, Briscoe B, Nemzer L. (1999)	Genetic Abortion: considerations for patient care	<i>J Perinat Neonat Nurs</i> 13(2): 47-59	Review of procedure options for abortions.		C	
Brambati B, et al. (1998)	First 10000 Chorionic Villus samplings performed on singleton pregnancies by a single operator.	<i>Prenat Diagn</i> 18: 255-266	1992-1995	10,000 chorionic villus to determine safety and accuracy of testing	3	
Brun J-L, et al. (2003).	Feasibility, accuracy and safety of chorionic villus sampling: a report of 10,471 cases.	<i>Prenat Diagn</i> 23(4): 295-301.	1990-1999	10,471 singleton pregnancies referred for CVS procedure to the fetal medicine unit of a French teaching hospital	3	Genetic testing diagnosis

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Biggio JR, et al. (2004)	An outcomes analysis of five prenatal screening strategies for trisomy 21 in women younger than 35 years.	<i>Am J Obstet Gynecol.</i> 190(3):721-9	Unreported	Hypothetical cohort of 1,000,000 women <35 years old	2	Cost-effectiveness analysis; genetic testing
California Birth Defects Monitoring Program (1995)	The National cost of Birth Defects	California Department of Health	1992	List of the cost of birth defects.	3	Birth defects, costs
Cardi van den Berg, et al. (1999)	Amniocentesis or chorionic villus sampling in multiple gestations? Experience with 500 cases.	<i>Prenat Diagn</i> 19: 234-244	1988-1997	500 women that underwent CVS and Amniocentesis to evaluate consequence of discordant results	3	Amniocentesis, cvs
Caughey A, et al. (2004).	Assessment of Demand for prenatal diagnostic testing using willingness to pay.	<i>Obstet Gynecol</i> 103(3): 539-45.	1997	447 women interviewed for willingness to pay for prenatal diagnostic services	2	Prenatal diagnostic testing
Center for Disease Control and Prevention (2003)	Births: Final Data for 2002.	<i>National Health Statistics Reports</i> , 52 (10)	Review: 2002 national birth data		A	Prenatal tests; utilization
Chilaka VN, et al. (2001).	Knowledge of Down syndrome in pregnant women from different ethnic groups.	<i>Pren Diagn.</i> 21: 159-64	Not reported.	245 pregnant British women of varying ethnic backgrounds	1	Prenatal testing; uptake of services
Ciarleglio LJ, et al. (2003).	Genetic counseling throughout the life cycle.	<i>J Clin Invest</i> 112(9): 1280	Review		A	Prenatal genetic counseling; prenatal genetic tests
Coleman KB (2002)	Genetic counseling in congenital heart disease.	<i>Critical Care Nursing Quarterly</i> 25 (3): 8-16.	Review		C	Genetic counseling
CORN and the Genetic Services Branch/MCHB/HRS A	Integrating Genetic Services into Managed Care.	Conference Presentation	Varying studies from different dates on genetic services.		A	Genetic services management (personnel communication with Doyle)

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Cunningham G and Tompkinson DG. (1999).	Cost and effectiveness of the California triple marker prenatal screening program	<i>Genet Med</i> 1(5): 199-206.	1995-1997	All pregnant Californians receiving prenatal care before 20 weeks gestation	3	Prenatal care; cost/benefit
DADA Study Group (2002).	Outcomes of Pregnancies diagnosed with Klinefelter syndrome the possible influence of health professionals	<i>Prenat Diagn</i> 22: 562-566.	1988-1997	Medical records of 111 women identified as having a fetus with KS	1	Prenatal diagnosis; counseling
Dormandy E, et al., (2002)	Variation in uptake of serum screening: the role of service delivery	<i>Prenatal Diagnosis</i> 22: 67-69	Not reported	29 hospitals in England	3	Screening uptake
Doyle, DL. (2003).	Health Care Consumers Cannot Benefit from the Advances in Medical Genetics Without Access to Knowledgeable and Qualified Genetic Service Providers.	Unpublished	Review		A	Geneti3 servi3es
Emery J, et al. (1999).	A systematic review of the literature exploring the role of primary care in genetic services.	<i>Family Practice</i> , 16: 426-45.	Review		A	Physi3i1n; geneti3 testing
Emery J, Hayflick S. (2001).	The challenge of integrating genetic medicine into primary care.	<i>BMJ</i> . 322: 1027-30.	Review		A	Physi3i1n; geneti3 testing
Eng C, et al. (1997)	Prenatal Genetic Carrier Testing using Triple Disease Screening	<i>JAMA</i> 278(15): 1268-1272	Not reported.	2824 Ashkenazi Jewish individuals referred for TSD testing	1	Prenatal genetic screening
Ensenauer RE, et al. (2003).	Primer on medical genomics part VIII: Essentials of medical genetics for the practicing physician.	<i>Mayo Clin Pro.</i> 78(7): 846	Review		A	3ost; geneti3 testing
Feldman B, et al., (2000)	Routine prenatal diagnosis of aneuploidy by FISH studies in high-risk pregnancies.	<i>Am J Med Genet</i> 90(3): 233-8.	1996-1998	FISH analysis of 301 pregnant women	3	Prenatal diagnosis

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Frisbie WP, Echevarria S, Hummer RA. (abstract only, 2001).	Prenatal care utilization among non-Hispanic Whites, African Americans, and Mexican Americans.	<i>Matern Child Health J.</i> 5(1): 21-33.	Unknown from abstract	Women of various ethnic groups in Texas.	1	Prenatal genetic testing.
Garber AM and Fenerty JP (1991)	Cost and benefits of prenatal screening for CF	<i>Med Care</i> 29(5): 473-487.	Not reported	Data on CF testing and selective abortion.	3	Genetic screening; cost of disease
Garel M, et al., (2002).	Ethical decision-making in prenatal diagnosis and termination of pregnancy: a qualitative survey among physicians and midwives.	<i>Prenat Diagn</i> 22: 811-7.	1999-2000	17 obstetricians and 30 midwives from three French maternity units.	3	Prenatal diagnosis
Genetics Education Center University of Kansas Medical Center.	Prevalence of Genetic Conditions/Birth Defects	Accessed online: www.kumc.edu	List of references for Genetic conditions		C	Geneti3 3onditions, 2irth defe3ts
Gibson AL, Doyle DL, Bryant S (2004).	Exploring the Educational Needs of Physicians: Do the Needs of Primary Care Providers Differ from Specialist Providers?	PowerPoint Presentation received from the authors	2004	Two focus group of Washington State providers	1	Challenges; genetic testing; physicians
Gollust DE, et al., (2003).	Direct-to-consumer sales of genetic services on the Internet.	<i>Genet Med</i> 5(4): 332-7.	Review: web-based sites offering genetic testing and services		A	133ess; geneti3 testing
Grobman WA, et al. (2002).	Preference assessment of prenatal diagnosis for Down syndrome: Is 35 a rational cutoff?	<i>Prenat Diagn.</i> 22: 1195-1200	1999-2000	186 women receiving antepartum care at a university hospital	1	Prenatal diagnosis; standard of care
Haddow JE, et al (1998).	Screening of Maternal Serum for Fetal Down's Syndrome in the First Trimester.	<i>NEJM.</i> 338 (14): 955-62.	1994-1996	Serum concentrations of five maternal analytes from 4,412 women with singleton pregnancies	2	Prenatal genetic screening

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Hall S, Abramsky L, Marteau M. (2003).	Health professionals' reports of information given to parents following the prenatal diagnosis of sex chromosome anomalies and outcomes of pregnancies: a pilot study	<i>Prenat Diagn</i> 23: 535-538.	Not reported.	29 health care professionals	1	Pregnancy outcomes, counseling
Hardy O, et al. (2003).	Hypothyroidism in Down Syndrome: Screening guidelines and testing methodology.	<i>Am J Med Genet.</i> 12A: 436-37.	Letter to the Editor.		C	Newborn screening guidelines.
Harris R, et al. (2004)	Cost utility of prenatal diagnosis and the risk-based threshold.	<i>Lancet</i> 363: 276-282	Not reported.	534 pregnant women who had CVS/amniocentesis or no invasive testing	2	Cost, Prenatal diagnosis
Himes P (1999)	Early pregnancy prenatal diagnostic testing: Risks associated with chorionic villus sampling and early amniocentesis and screening options.	<i>J Perinat Neonat Nurs</i> 13(2): 1-14	Review		C	Prenatal diagnostic testing.
Holems-Siedle M, Ryyanen M, Lindenbaum RH. (1997)	Parental decisions regarding termination of pregnancy following prenatal detection of sex chromosome abnormality.	<i>Prenat Diagn</i> 7(4): 239-244.	1970-1984	Forty cases where fetus were identified as having a genetic abnormality about terminations of pregnancy	1	Pregnancy terminations, prenatal genetics
Hollander A., (2004).	Defining Genetic Information and Genetic Tests: A Comparison of State Statutes.	Available online at: http://www.genelaw.info	Not reported	State statutes related to genetics in healthcare.	2	Genetic test
Institute for Clinical Systems Improvement (2003).	Routine prenatal care.	Accessed online from the National Guideline Clearinghouse of the Agency for Health Care Research and Quality (AHRQ) www.guideline.gov	Clinical guidelines for the practice of routine prenatal care		B	Prenatal diagnosis; practice guidelines

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Jaques A and Sheffield L (2003).	Letter to the Editor: Uptake of prenatal screening for chromosomal anomalies: impact of test results in a previous pregnancy.	<i>Prenat Diagn</i> 23: 1097-1103		Letter to the Editor: suggest high statistical significance exists in prenatal screening uptake rates in women of low-risk as compared to high-risk groups.	B	Dem1nd; prenatal genetic testing
Khoshnood B, et al. (2004).	Maternal education modifies the age-related increase in the birth prevalence of Down syndrome.	<i>Prenat Diagn.</i> 24: 79-82.	1989-1991	Data from the national birth statistics database (n=8,319,939 births)	1	Utilization; prenatal diagnosis; testing
Koscica KL, et al. (2001).	Assessing genetic risk: Comparison between the referring obstetrician and genetic counselor.	<i>Am J Obstet Gynecol.</i> 185(5): 1032-34.	1999	Charts from 145 patients referred for genetic counseling	1	Genetic screening.
Kubas C. (1999).	Noninvasive means of identifying fetuses with possible Down syndrome: a review.	<i>J Perinat Neonat Nurs</i> 13(2): 27-46.	Review		B	Prenatal diagnosis, testing
Lawson K (2003).	Perceptions of deservedness of social aid as a function of prenatal diagnostic testing.	<i>Journal of Applied Social Psychology</i> 33(1): 76-90.	Not reported.	341 physicians and 281 university employees in Canada	3	Prenatal diagnostic testing
Leithner K, et al., (abstract only, 2004)	Affective state of women following a prenatal diagnosis: predictors of a negative psychological outcome.	<i>Ultrasound Obstet Gynecol</i> 23(3): 240-6	Unknown from abstract	77 pregnant women undergoing prenatal diagnostic procedures.	2	Outcome genetic testing; prenatal diagnosis
Linden MG, Bender BG, Robinson A. (2002)	Genetic Counseling for Sex Chromosome Abnormalities	<i>Am J Med Genet</i> 110: 3-10.	Review: follow-up of 307 individuals born with sex chromosome abnormalities between 1964-1975.		C	Counseling; prenatal diagnosis
Mansfield C, et al. (1999).	Termination Rates After Prenatal Diagnosis of Down Syndrome, Spina Bifida, Anencephaly, and Turner, and Klinefelter Syndromes: a systematic literature review.	<i>Prenat Diagn,</i> 19: 808-812	Review		A	Prenatal diagnosis, termination

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Marteau TM. (2002)	Prenatal testing: towards realistic expectations of patients, providers and policy makers	Ultrasound Obstet Gynecol 19: 5-6	Review		B	Prenatal testing
McCandless SE, Brunger JW, and Cassidy SB (2004).	The burden of genetic disease on impact care in a children's hospital	<i>Am J Hum. Genet</i> 74 121-127.	1996	5747 hospital admissions at Rainbow Babies and Children's Hospital to estimate cost of care for genetic illness	3	Genetic disease, burden
Meschede D, Albersmann S, and Horst J. (2000).	The practical importance of pedigree analysis in women considering invasive prenatal diagnosis for advanced maternal age or abnormal serum screening tests.	<i>Prenat Diagn</i> 20: 865-9.	Not reported	1,356 pregnancies in Germany in which the either: 1) the mother was older than 35 years; 2) there was an increased risk for Down syndrome based on triple screen results; 3) there was an increased risk for NTD based on maternal AFP screening results.	3	Prenatal diagnosis; genetic counseling
Michie S, et al. (1997)	Nondirectiveness in genetic counseling: an empirical study	<i>Am J Hum Genet</i> 60(1): 40-47	Not reported.	131 transcripts of genetic counseling session and reports from counselors and counsees about nondirectiveness	1	Genetic counseling
Michie S, et al. (2002).	Predictive Genetic Testing: High Risk Expectations in the face of low risk information.	<i>Journal of Behavioral Medicine</i> 25(1)33-50.	Not reported	127 unaffected adults at risk for FAP in the UK assessed for expectation of screening for FAP	3	Genetic screening
Michie S, Marteau TM, Bobrow M. (1997)	Genetic Counseling: the psychological impact of meeting patients' expectations	<i>J Med Genet</i> 34(3): 237-41	Not reported	131 individuals that participate in genetic counseling in the UK.	1	Genetic counseling outcomes
Michie S, McDonald V, Marteau TM (1997)	Genetic counseling: information given, recall and satisfaction	<i>Patient Edu. Couns.</i> 32(1-2): 101-106	Not reported	32 individuals in the UK that participated in genetic counseling questioned	1	Genetic counseling outcomes

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Muller F, et al. (2002).	Risk of amniocentesis in women screening positive of Down syndrome with second trimester maternal serum markers.	<i>Prenat Diagn</i> 22: 1036-9.	1997-1999	50,476 patients who had maternal serum screen analysis performed by one of six French laboratories.	3	Prenatal diagnosis; amniocentesis
Murthy V. (2000).	Why Aren't Women Utilizing Prenatal Genetic Services in Washington State? – What Genetic Counselors Think.	Unpublished	2000	18 genetic counselors in Washington State	1	Barriers to genetic services; genetic counseling
National Coalition for Health Professional Education in Genetics (2003).	The Genetic Family Tree: Not Just for Geneticists.	<i>In Practice</i> 1(1): 1-5.	Review. The importance of pedigree analysis in assessment of genetic risk		A	Genetic services; risk assessment
National Society of Genetic Counselors	Professional Status Survey.	National Society of Genetic Counselors, Inc. Accessed online from http://nsgc.org	2002	856 members of the NSGC.	1	Genetic counseling; counselor
National Society of Genetic Counselors	What is Genetic Counseling?	Received from DL Doyle, 2004.	Patient-oriented education pamphlet		A	Genetic counseling
Noorani, et al. (1996).	Cost Comparison of Molecular versus Conventional Screening of relatives at Risk for Retinoblastoma	<i>Am J. Hum Genet.</i> 59 301-307.			2	Costs of screening
Papp C. (2003).	Chorionic Villus Sampling.	<i>Ultrasound Review of Obstetrics and Gynecology</i> 3(4): 279-85.	Review		C	Prenatal diagnosis
Petrou S and Henderson J.(2003)	Preference-based approaches to measuring the benefits of perinatal care	<i>Birth</i> 30 (4) 217-225	Review		A	Prenatal care; costs and benefits
Pettersen B.	Summary of Billing/Reimbursement Surveys	Accessed online: http://nsgc.org	2002	14 Genetic Counselors in United States	1	Billing genetic counseling

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Pettersen B., with contributions from various sources (2003).	Primer on Billing and Reimbursement for genetic counselors	NSGC Genetic Services Committee	Review		A	2illing geneti3 3ounseling
Press N and Browner CH. (1998)	Characteristics of Women Who Refuse an Offer of Prenatal Diagnosis: Data from the California Maternal Serum Alpha Fetoprotein Blood Test Experience.	American Journal of Medical Genetics. 78: 433-45.	1989-1992	Pregnant women who declined MSAFP screening at one of five sites in Southern California (all run by the same HMO).	1	Prenatal diagnosis; screening; utilization
Rapp R. (1994).	<u>Women's Responses to Prenatal Diagnosis: A Sociocultural Perspective on Diversity.</u>	In <i>Women and Prenatal Testing: Facing the Challenges of Genetic Technology</i> (K. Rothenberg and E. Thomson, eds.) Ohio State University Press (1994) pp 219-233.			1	Prenatal genetic testing; public response
Raymond MZ (2003)	Current Trends in Prenatal Genetics: What the Data Tells Us.	(unpublished)	1997-2002	All women seen for genetic services at one of ten Washington State Regional Genetics Clinics	1	Prenatal genetic services; utilization
Raymond MZ (2004).	The Availability of Prenatal Genetic Services in Washington State	Master's Thesis. University of Washington School of Public Health and Community Medicine.	1998-2002, 2004	Statewide pregnancy and birth statistics; 1959 state-licensed obstetric providers	1	Prenatal genetic services; availability; obstetric providers
Roberts CD, Stough LM, Parrish LH. (2002).	The Role of Genetic Counseling in the Elective Termination of Pregnancies Involving Fetuses with Disabilities.	<i>Journal of Special Education.</i> 36(1): 48-55	Not reported.	69 women who, following prenatal screening, were at increased-risk for carrying a fetus with a disability.	1	Prenatal diagnosis; genetic counseling
Robinson A, Bender BG, Linden MG. (1989)	Decisions following the intrauterine diagnosis of sex chromosome aneuploidy.	<i>Am J Med Genet.</i> 34(4): 552-4.	Not reported	327 parents that had a prenatal diagnosis of SCA decisions about termination	1	SCA, terminations

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Robinson A, Bender BG, Linden MG. (1992).	Prognosis of prenatally diagnosed children with sex chromosome aneuploidy.	<i>Am J Med Genet</i> 44(3): 365-368.	Not reported	530 parents that received prenatal diagnosis of SCA	3	Prenatal diagnosis
Roizen NJ and Patterson D. (2003)	Down's syndrome.	<i>Lancet</i> 361 (9354): 1281	Review		C	Prenatal diagnosis
Rollnick B (1984).	A time analysis of a genetic counseling delivery system	<i>Birth Defects Orig Artic Ser.</i> 20(6):208-12.			3	Strategies in Genetic counseling; clinical investigation studies.
Russel PJ (1998).	Genetics (5 th edition)	Menlo Park, CA: Benjamin/Cummings Publishing Co, Inc.	College-level introductory genetics textbook		A	
Schoonmaker MM, Bernhardt BA, Holtzman NA (2000).	Factors influencing health insurers' decision to cover new genetic technologies.	<i>Int J Technol Assess Health Care.</i> 16(1):178-89.	Not reported	National survey	1	Payment; genetic screening
Sinclair A. (2002).	Genetics 101: Cytogenetics and FISH.	<i>Journal of the Canadian Medical Association.</i> 167(4): 373	Review		C	Genetic testing; diagnosis
Singer E, Antonucci T, Van Hoewyk J. (abstract only, 2004).	Racial and ethnic variations in the knowledge and attitudes about genetic testing.	<i>Genet Test</i> 8(1): 31-43.	Unknown from abstract	Telephone survey of African-Americans, Latinos, and non-Hispanic Whites	1	Genetic testing.
Tanski S, Shulam Rosengren S, and Benn PA (1999)	Predictive Value of the Triple Screening Test for the Phenotype of Down Syndrome.	<i>Am J Med Genet</i> 85: 123-6.	1991-1997	30 cases of child born with Down syndrome cases	3	Prenatal diagnosis; maternal serum screening
Tepperberg J, et al., (2001).	Prenatal diagnosis using interphase fluorescence <i>in situ</i> hybridization (FISH): 2-year multi-center retrospective study and review of the literature.	<i>Prenat Diagn</i> 21: 293-201.	1998-1999	The authors collected FISH analysis and karyotypes of 5,348 pregnancies from 25 U.S. laboratories	3	Prenatal diagnosis

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Tercyak K, et al. (2001)	Psychological response to prenatal genetic counseling and amniocentesis	<i>Patient Education and Counseling</i> 43: 73-84	Not reported.	129 individuals referred to genetic counseling in a southeastern hospital	1	Prenatal testing, Genetic counseling
Waitzman NJ, Romano PS, Scheffler RM. (1994)	Estimates of the Economic Costs of Birth Defects	<i>Inquiry</i> 33: 188-205.	Not reported	Cost estimates of 18 clinically significant birth defects	1	Cost; birth defects
Wald NJ, et al., (2003)	Cystic fibrosis: selecting the screening strategy of choice.	<i>Prenat Diagn</i> 23: 474-83.	Review		A	Prenatal genetic screening
Wang C, Gonzalez R, Merajver SD. (2004).	Assessment of genetic testing and related counseling services: current research and future directions.	<i>Social Science and Medicine</i> 58: 1427-42.	Review of research in genetic services.		A	Genetic counseling; outcomes; testing
Washington State Department of Health Community and Family Health and Department of Social Health Services (2000).	Prenatal Diagnosis Genetic Counseling: Billing Instructions	Received from DL Doyle, Washington State Department of Health	Billing instructions		A	Genetic counseling; prenatal billing
Washington State Department of Health Community and Family Health and Department of Social Health Services (2004).	Prenatal Diagnosis Genetic Counseling: Fee Schedule Changes	Accessed online: http://mma.dshs.gov.wa	Updates to previous codes		A	Genetic counseling; prenatal billing
Washington State Department of Health, (2002).	Pacific Northwest Laboratory Medicine Sentinel Monitoring Network – Final Report of the Findings of Questionnaire 17: Molecular genetic testing.	Received from DL Doyle, Washington State Department of Health	2002	204 “high complexity laboratory network participants”.	2	Genetic testing

Author(s)	Title	Journal	Study Date	Population Studied	Relevance	Key Words
Wilkins-Haug, et al., (1999).	Genetics in Obstetricians' Offices: A Survey Study.	<i>Obstet Gynecol</i> 93: 642-7.	1997	446 ACOG fellows practicing obstetrics	1	Physician and prenatal genetics
Wille MC, et al., (2004).	Advances in Preconception Genetic Counseling.	<i>J Perinat Neonat Nurs</i> 18(1): 28-40.	Review		A	Preconception genetic counseling;
Willner JP. (1998).	Reproductive Genetics and Today's Patient Options: Prenatal Diagnosis.	<i>Mount Sinai Journal of Medicine.</i> 65(3): 173-77.	Review		A	Genetic testing; screening; prenatal diagnosis
Zoppi MA, et al. (2001)	Nuchal Translucency and the Acceptance of Invasive Prenatal Chromosomal Diagnosis in Women Aged 35 and Older.	<i>Obstet Gynecol</i> 97: 916-20.	1995, 1999	Two cohorts of women of advanced maternal age, referred for prenatal diagnosis in either 1995 or 1999.	3	Prenatal diagnosis

APPENDIX C

Summary of Secretary's Advisory Committee on Genetics, Health, and Society Draft Report "Coverage and Reimbursement of Genetic Tests and Services"

The draft report titled, "Coverage and Reimbursement of Genetic Tests and Services," issued by the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) describes the current state of, and problems associated with, coverage and reimbursement of genetic tests and services. It also proposes recommendations to the Secretary of Health and Human Services on ways to improve current coverage and reimbursement mechanisms to provide for appropriate access to and utilization of health-related genetic tests and services.

The report proposes the following definition for 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, and genetic counselors. They include:

- *Laboratory services that involve the provision of tests using genetic/genomic technologies, interpretation of results, and oversight of the test's performance;*
- *Identification or diagnosis of individuals and families at risk for a disorder with a genetic component or who could benefit from pharmacogenomic testing; and*
- *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.*

The Committee notes that there are several challenges to the integration of genetic tests and services. These include the same challenges that other new technologies face as well as unique challenges such as those related to the coordination of care involving multiple providers, complex gene environment interactions, the concept of diagnosis versus identification of a risk factor, and the implications genetic test results may have for blood relatives.

The report examines methods, including evidence-based methods, that private and public health insurance providers use to make decisions about which services are going to be covered by their plans. These include technology assessments, assessing informational utility, and cost-effectiveness analyses. Other factors that influence coverage decisions are legal requirements, consumer demand, and whether the service is covered under other plans such as Medicare.

Medicare coverage decisions can be made at the national or local level. If the national coverage determination policy is to exclude a test or service from coverage, then the local Medicare contractor may not cover the service either. However, in the absence of a national coverage determination, local contractors can make a local coverage determination. In order to be considered for coverage under the Medicare plan, tests and services must meet specific criteria; they must fall within the scope of benefits as defined by Congress, be approved by the Food and Drug Administration (FDA), and meet a "reasonable and necessary" requirement. In general, Medicare does not cover screening services.

Medicaid coverage decisions are also made at the national and local levels. The federal government mandates that certain benefits be provided, but states have flexibility to cover additional benefits; some additional benefits require the use of state funds without matching federal dollars. Differences in Medicaid coverage result in disparate access to genetic tests and services by state.

The report describes how health care providers bill third party payers for services and how those payers reimburse providers. Coding systems have been developed and are used to describe causes of morbidity, treatments, and time spent with patients. The coding systems are updated periodically to account for changes in available services, but these updates may not keep pace with rapid changes in knowledge and technology. The codes are inadequate for describing many genetic tests and services and reimbursement rates are often below the actual cost of providing the services. These inadequacies are especially apparent with respect to laboratory services and genetic counseling services.

The report ends with a brief discussion of three related issues. The Committee identified health disparities, education and training of providers, and public awareness as issues that affect how information is gathered and disseminated and therefore have an effect on coverage decisions.

The draft report includes the following potential recommendations:

1. The Secretary should task an appropriate group or body to develop a set of principles to guide coverage decision making for genetic tests. The principles should identify criteria to help determine which types or categories of genetic tests should be covered, which should not be covered, and which fall into an uncertain gray zone. The group's guiding principles should address the issues of economic evaluation/cost-effectiveness, prevention, rare disease tests, and therapeutic versus informational benefit. The Committee also recommends that the existing evidence for specific tests be assessed in order to determine whether the evidence is adequate in type, quality, and quantity to establish analytical validity, clinical validity and clinical utility as well as to identify any gaps in evidence.

This body should include both relevant HHS agencies and private sector organizations and utilize resources or models in the public and private sector. The EGAPP Work Group (see box below) involves such a diverse range of stakeholders and is performing similar work and, thus, is an example of such a body to be tasked to develop these principles and address these issues.

The Committee also recommends a mechanism be established that would specifically promote and fund studies to address any identified gaps in the evidence base.

2. Genetic tests and services in pediatrics and those with a prevention component should be considered specifically with respect to the benefits they can offer the populations they serve. Although standardization of coverage decisions using best scientific evidence across public and private payers is ideal (see Recommendation 1), private payers should be supported with necessary information to make their own coverage determinations about these tests and services relative to the populations they serve.

3. Although a mixed national-local coverage decision-making process is a reasonable approach to making Medicare coverage decisions for genetic tests and services, there are several aspects of the current national-local decision-making process that limit its utility. While not suggesting changes to the current system, SACGHS recommends that the Secretary encourage the Centers for Medicare and Medicaid Services (CMS) to move forward with the implementation of Section 731 of the Medicare Prescription Drug, Improvement, and Modernization Act of 2003, which requires 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.

4. Medicare beneficiaries who lack current signs, symptoms, or personal histories of illness stand to benefit clinically from predictive and predispositional genetic tests and services. As such, SACGHS recommends that preventive services, including predispositional genetic tests and services, meeting 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 determine through its national coverage decision-making process (which includes an assessment of existing evidence) whether an item or service is reasonable and necessary for the prevention or early detection of an illness or disability and, thus, ought to be covered. Such action would allow CMS to consider covering many more genetic tests and services that are used for preventive purposes.

More immediately, the Secretary should direct CMS to clarify, through appropriate guidance, that in certain circumstances and where scientific evidence warrants, "personal history" may include family history of a particular disease for purposes of establishing that a genetic test is "reasonable and necessary" and, therefore, covered under Medicare. CMS should specify the circumstances and criteria required to make such a determination.

5. The Secretary should broadly disseminate to all states information about the existing evidence base and other supporting information, such as guiding principles that serve as the basis for coverage decision making on genetic tests and services. This information could be used by states to inform their Medicaid coverage decisions.

HHS should continue to provide states with grants that encourage the coverage, adoption and provision of genetic services that have a sound evidence base.

6. In many cases, payment rates for genetic tests are lower than the actual cost of performing the test. Until the fee schedule can be reconsidered in a comprehensive way, the Secretary should direct CMS to address variations in

payment rates for the genetic test Current Procedural Terminology (CPT) codes through its inherent reasonableness authority.

7. Genetic counseling is a critically important component of the appropriate use and integration of genetic tests and services. As such, SACGHS recommends the following:

- Qualified health providers should be allowed to bill directly for genetic counseling services. The Secretary should expeditiously identify an appropriate mechanism for determining the credentials and criteria needed for a health provider to be deemed qualified to provide genetic counseling services and eligible to bill directly for them.
- The Secretary should direct government programs to reimburse prolonged service codes when determined to be reasonable and necessary.
- HHS, with input from the various providers of genetic counseling services, should assess the adequacy of existing CPT evaluation and management (E&M) codes and their associated relative values with respect to genetic counseling services. Any inadequacies identified should be addressed as deemed appropriate.
- CMS should deem all non-physician health providers who are currently permitted to bill directly any health plan – public or private – eligible for a National Provider Identifier (NPI).
- The Secretary should direct CMS to allow non-physician health professionals who are qualified to provide genetic counseling services and who currently bill incident to a physician to utilize the full range of CPT E&M codes available for genetic counseling services.

8. Since providers act as intermediaries between health plans and plan members and thus have an important role in ensuring genetic tests and services are provided appropriately, there is a need to support the ongoing training and continued education of health providers in genetics and genomics. SACGHS's recommendations to the Secretary in 2004 included the following: the Secretary should develop a plan for HHS agencies to work collaboratively with state, federal and private organizations to support the development, cataloguing and dissemination of case studies and practice models that demonstrate the current relevance of genetics and genomics; and the Secretary should strive to incorporate genetics and genomics into relevant initiatives of HHS, including the National Health Information Infrastructure.

9. Reliable and trustworthy information about family history, genetics and genetic technologies should be developed and made more widely available through the internet and other mechanisms that allow patients and consumers to evaluate health plan benefits and health providers so that they may make the most appropriate and most financially responsible decisions for themselves and their families.

The Secretary should leverage HHS resources to develop and make widely available reliable and trustworthy information about family history, genetics, and genetic technologies to guide and promote informed decision making by healthcare consumers and providers. Such information should be made available information through federal government websites and other appropriate mechanisms.

May 5, 2005

Reed V. Tuckson, M.D.
SACGHS Chair
Secretary's Advisory Committee on Genetics, Health, and Society
Department of Health and Human Services
6705 Rockledge Drive
Suite 750, MSC 7985
Bethesda, MD 20892-7985

Dear Dr. Tuckson:

Thank you for the opportunity to review and comment on the draft report titled, "Coverage and Reimbursement of Genetic Tests and Services." On behalf of the Genetic Services Policy Project (GSPP), I would like to commend the committee members and staff for addressing this important issue in the delivery of genetic services. I would also like to offer a few comments based on our experience in the GSPP.

As the report cogently points out, genetic services have great potential for improving health for many people. However, in the current fiscal environment, increasing access to genetic services means decreasing access to other beneficial services. When resources are scarce, effectiveness is a necessary but not sufficient condition for coverage. The new service competes for dollars with other health care services that are also effective (although perhaps for other conditions and/or other people). In addition, dollars spent on health care, particularly public health care dollars, compete with dollars spent on other (non-health care) services of value. Thus, as with many new medical technologies, expanding coverage for genetic services should proceed thoughtfully and with consideration for competing demands on resources.

Determining the right trade-off among alternative services that have potential health benefit, particularly when the distribution of benefits across different populations is uneven, necessarily involves value judgments. The committee's report and proposed recommendations state very clearly the values and perspectives of genetic services providers. This is certainly an important perspective. However, there are other perspectives that are not reflected in the report. In our experience working with a diverse advisory committee representing providers, business, payers (public and private), consumers, and policy makers, we have found that these groups often have different views. In our opinion, considering many perspectives on the issues central to genetic services financing and delivery is not only useful but also provides the appropriate balancing of science and values.

Overall, the draft report is a well-written, thoughtful description and analysis of coverage and reimbursement issues as related to genetic tests and services. Thank you for giving your attention to this issue.

Sincerely,
Carolyn (Cindy) Watts, Ph.D.
Professor
Genetic Services Policy Project

GSPP is a federally-funded collaborative effort between the University of Washington, the Washington State Department of Health, and the Fred Hutchinson Cancer Research Center. The purpose of the project is to describe the current genetic services delivery system; collect, develop, and disseminate information to stakeholders; and to propose alternative genetic services delivery models through consultation with a diverse advisory committee, which comprises representatives from various key stakeholder groups. GSPP is supported by projects # U35MC02601 and # U35MC02602 from the Maternal and Child Health Bureau (Title V, Social Security Act), #11223, Health Resources and Services Administration, Department of Health and Human Services.

APPENDIX D

HRSA Project Advisory Panel Meeting The Churchill Hotel, Washington DC, September 24, 2004 AGENDA

8:00 a.m.

Morning Refreshments

8:15 a.m.

HRSA Project Management Introductions

Greetings and thanks from HRSA, Statement of HRSA objectives and expectations, Introduction of the Genetic Services Policy Project Team

8:45 a.m.

Orientation

Around the room introductions by Advisors, each taking 5 minutes to describe their role in the field and within their organization, their particular expertise and/or professional interests, and a brief statement of their expectations for the project.

10:00 a.m.

Break

10:30 a.m.

Discussion of HRSA Project Conceptual Framework

11:00 a.m.

Genetic Services Elements

Facilitated discussion of the elements constituting genetic services, presentation of the provisional classification system for genetic services, and solicitation of recommendations from Advisors on modifications, additions, etc. to the scheme, particularly in light of Project 4 requirements.

12:00 p.m.

Break for check-out, etc.

12:15 p.m.

Working Lunch

Presentation and discussion of enabling technologies for users, (e.g. EMRs, MCO member service interfaces such as Vivius), and discussion of genetic testing services, including direct-to consumer services.

1:45 p.m.

Breakouts

Discussion about What information Advisors want about genetics; How to receive this information; When; from Whom; and How will it be used?

2:45 p.m.

Break

3:00 p.m.

Reporting back by groups, and discussion

3:30 p.m.

Feedback and suggestions from Advisors on both content and process, and next steps,

4:00 p.m.

Adjourn

Genetic Services Policy Project (GSPP) Advisory Panel Meeting
Meeting Minutes
September 24, 2004

The first meeting of the Genetic Services Policy Project Advisory Panel Meeting was held at the Churchill Hotel, Washington DC on September 24, 2004. Present were:

Advisors:

Jean Anderson
Meg Booth
Daragh Conrad
Brett Davis
Patricia Deverka
Sarah Donta
Donald Kemper
Joseph McInerney
Lee Newcomer
Lawrence O'Connor
Daniel Perry
Barry Steinhardt
Peggy Stupca
Sharon Terry
Brad Therrell
David Weismiller
Peter West
Mark Yarborough

Project Staff and Federal Partners:

Wylie Burke
Rick Carlson
Debra Lochner Doyle
Penny Kyler
Michele Lloyd-Puryear
Merle McPherson
Amber Roche
Sherry Taylor
Cindy Watts

Consultants:

Leslie Wainwright

The following represents a brief overview of discussions held and is not a detailed and complete transcript.

Genetic Services Policy Project
Management Introductions

HRSA Introductions

Penny Kyler, HRSA Project Officer

Overview (see attached handouts) of where the project fits within Health Resources and Services Administration (HRSA). Maternal and Child Health Bureau, Division of Services for Children with Special Health Needs, Genetic Services Branch.

Genetic Service and Health Care Delivery, is also looking at the cross-cutting issues that are disease specific or systems specific and how they can come together.

The GSPP is nested within the concept of Translational Genetics. Also, within Translational Genetics HRSA is looking at the Public Policy Component as well as the educational component capacity for analysis and applied research projects.

Michele Lloyd-Puryear, HRSA Chief, Genetic Services Branch

These are two very important projects. Your work here will lay out HRSA's future work around providing national leadership for a broad population. HRSA is a service organization so the goals of these projects will be in the area of "services" and community based education. This Project is a cooperative agreement – a partnership between the project's staff and HRSA, where we both bring resources to the table.

Merle McPherson, Division Director, Office of Children with Special Health Care Needs

The Genetics part of this initiative speaks to a huge emerging field that has wonderful new science to it. Much of the work there is translational science.

The mandate we now have, is a family-centered comprehensive care approach. This portion is very much an implementation and sustainability. It is not a demonstration.

ACTION: none

Orientation Around the table introductions by Advisors.

ACTION: none

Discussion of Genetic Services Policy Project Elements **Wylie Burke**

Overview (see attached handouts) of Genetics, Medicine, and Public Health. Diverse applications of genetics with diverse challenges in terms of bringing good services to people, in terms of the necessary professional and public education, and in terms ethical, legal, and social implications.

We face a number of questions: When does harm of a genetic label outweigh its benefit? Who decides when new tests and interventions are ready for use, and on what basis? And, how do we ensure access to all who can benefit?

Debra Lochner Doyle

The success of this project is determined by the experience shared by our diverse group of advisors, and the partnership between HRSA, the Washington State Department of Health, the University of Washington, and the Fred Hutchinson Cancer Research Center. It is very much a collaborative effort.

How are genetics services delivered, particularly in states with a poorer genetic services provider base. Are there alternative ways to deliver services in those areas, such as telemedicine, etc.

What types of educational materials might work best for your constituency groups, what information do they need or want, and how do they want that material disseminated?

Cindy Watts

Overview (see attached handouts) of the economics of the project in terms of who uses genetic services and why; who delivers the services and where; who pays for the services, and how do the providers get paid? Where do consumers and providers get their information about services? What is the government's role? What are the cultural, ethical and policy issues? What are alternative delivery models, and, what public policy changes do we need?

ACTION: none

Genetic Services Policy Project Elements

Rick Carlson

Overview (see attached handouts) of the changes coming in healthcare in the larger context. Genomics adds new knowledge every day. How do we deal with the new knowledge it provides? If we get into a consumer market, we will begin to see people make their own healthcare decisions and medical services. If we give individuals that ability (to decide for themselves), we have to be prepared to have them make decisions that we (as healthcare providers) will think are wrong. We have to honor that decision unless it could result in great harm.

ACTION: none

Lunch Presentations

Lee Newcomer – Vivius

Consumer involvement in self-payment in health care is dramatically increasing, spurring the need for transparency of information to consumers so they can make

more informed and better decisions.

Consumer choice is simply another name for cost shifting. Consumers are currently paying 48% of their out-of-pocket health care expenses.

Pilot program in Spokane to enable consumers to determine who their healthcare providers were, with their associated referring physicians. Premiums were set based on who the mix of physicians consisted of. Preconfigured packages of healthcare providers with its own unique price. These packages could be custom built to cover specific health-care needs, with its resultant price increase.

Insurance industry not interested in this model. Employers and consumers love this model. The close ratio was ~60%.

Brett Davis – IBM

Overview of information based medicine and why we see information technology as being so critical to the future of healthcare. Background of project with Mayo Clinic, at the cutting edge of IT within healthcare.

Dr.s today are diagnosing based on experience. At the genomics level you are not going to be able to memorize everything. It becomes critical to know how to access information to make diagnosis. We define Information-based medicine as the process of improving the existing pharmaceutical and medical practice and the knowledge generated from the integration of diverse clinical and medical data.

Lots of data are being captured electronically in silos throughout healthcare. More and more data is being generated as new technology comes on board and as new tests and research increase healthcare opportunities. There are volumes of environmental data out there that are also siloed and often not integrated with genotypic and phenotypic information.

Data can be captured, integrated, and used in three different capacities. One being basic research, also in the clinical research and development, and finally in clinical delivery. Can revolutionize the way disease is diagnosed and treated.

Mayo Project takes clinical data and integrates it with the research data that has been generated at the Mayo Clinic, and also with external and public databases that exist in that have emerged in the last 10 or 15 years. (such as disease registries)

All the information about disease is out there, its just not easy to query. Phase I integrated the data, over 4.4 million patient records, in a way that clinicians could query it. Prior to integration a query would take approximately 6 weeks with 4 people devoted full time to accessing various databases for the data. After phase I the query took 6 seconds to answer. Pretty big impact on productivity. Being used in the research environment currently but expected to be used ultimately in the clinical setting with direct patient care.

Phase II integrates the research data and allows unstructured text searches.

ACTION: none

Breakout Sessions Group One Summary:

We started off with a fairly lively discussion about how we might communicate with this group over the next year until we are able to meet again face-to-face. One of the things that we agreed upon was that the BLOG was the right way to do our routine communications. We will be parsimonious in our requests of you, we will make specific requests, we will not give solutions, but questions to take the most advantage of your expertise. We might also use web-casts and conference calls and not use list-serves to clog up your email. Also, we will be religious about giving you progress reports on the project.

Themes:

There are some cross-cutting issues that cut across all of the themes that kept reoccurring.

Its really important that we look at all of this from various perspectives, that we aren't always focusing on the provider perspective or the payer perspective or any other perspective. One of the strengths of this group is that we are going to look at all sorts of different perspectives and we need to keep that in mind. We need to stay cognizant of the regulatory context and the impact of existing regulations on service delivery and secondly we need to be aware of the different business models that different stakeholders have from which they operate.

The next theme was about information This is clearly a very important theme throughout the project. Who gets it? Who controls it? How do we manage it? Does it matter who pays for it, in terms of the answers to any of those questions?

A related issue is value determination, and whose values count in making decisions about all of the above?

The last theme is the issue of consumer driven healthcare and its implications.

Ideas for the Case studies to be developed:

- A case study around who would benefit from a specific drug.
- A case involving the criteria used to evaluate a new genetic service.
- A case in which the person being tested doesn't really expect to take action (predetermination of disease).
- A case study that involves the duty to warn and what constitutes adequate discharge of that duty: the issues around liability.
- A case study of a condition involving adult onset of a genetic disease where there were some primary prevention activities that could be taken in adolescence to prevent or mitigate the impact of the disorder.
- Newborn screening –What can we learn from the newborn screening experience that would translate to other areas?
- Coverage and Universal Coverage. It might be interesting to look at an international example in a country where there is universal coverage. Look at the interaction between universal coverage, the coverage and utilization of preventive services and genetic services in particular. When there is universal coverage, there is more emphasis on evidence based decision making, typically around benefits for a cohort rather than an individual.
- A case study that would have us deal with the practicality of genetic services provided in clinical offices where clinicians have limitations on their time and resources.
- Concept of beneficence: value determination. Whose values? Is it enough that this test will give the consumer information? Even if obsess/he doesn't behave differently, is there still value in the information? Who gets to decide? Perhaps the behavior change is not readily observable. Is the answer dependent on who pays for the test?
- Does testing affect the availability of insurance? Insurance companies are already making coverage decisions based on family history. One of the ways in which genetic information might actually increase a person's ability to get insurance coverage might be if a consumer has a family history of breast cancer (and so might be heavily underwritten), and then tests negative for the BRCA1 mutation..

providers and others manage huge volume of new information on all patients.

Ideas for the Case studies to be developed:

Who will benefit from drug, how to manage within complex system.

- affect patient?
- provider?
- manufacturer?
- overall economics?

Regulatory environment – some important information not observable – need to have hypothetical too.

Drug company will/may suppress information about value

Payer will define context within which reimbursable

Value determination

- who's values?
- informed by whom?

What criteria are used by various stakeholders to evaluate new genetic services?

Need to consider business models

Predetermination of disease when “consumer knows s/he won't act on information

Should payment for test be predicated on use of information?

Who pays for beneficence?

- family centered care – decision maker
- value of information – who's value?

Medical model vs family/broader model

Public health/private health

Duty to warn/ defensive medicine

JAMA OB Literature

Genetic test as defensive medicine

Trade association's policy on existing statements

What constitutes adequately discharging duty to warn?

Can we get providers and patients on same page around right information?

Primary prevention around adult onset genetic diseases (major chronic disease)

Create hypothetical that involves drug and treatment.

What is health? What is illness? The unpatient

What does genetic literacy mean (providers, patients)

Need different kinds of information for providers vs patients

Goal – health promotion – eliminate all sources of disease except genetic factors.

Genetics drives medicine information science of difference.

Difference between what is beneficial and what we can afford.

Bad at doing tech assessment

Consumer choice is ultimate of value?

Flow chart of d-m by different stakeholders.

Who should have control over information?

Individual medical record

Should information go only to medical home?

Does concept of medical home have legs outside newborn screening?

What we do to our infants is different from what we do to adults.

NBS system provides lots of experience with a specific context

Application of child IQ information/ HIV information

Case study on right to information – Lee's case information only

Does right to know attach to who pays?

Nothing about me without me

Patients see value in information

Payers see little value in inactionable information

Universal/single payer devalues personalized medicine

Case study: international example – is universal coverage compatible with preventive genetic information.

Case – what is practicality of genetic service with limits on clinician's time and money?

Education – evidence-based trend moves focus to cohort not individual

Not blasts of fragments, threaded discussions – on website or bulletin board

Boss can summarize and make specific requests and trigger response
Library of resources
Blog with passwords
NOT listserves
Make individual requests
Conference call/ webcast
Genetic Alliance is developing survey tool with immediate feedback and links to more information.
Quarterly update on project – progress reports
One week response time
Come to us with questions vs solutions
Periodic sub-groups around specific issues
Distribute notes.

ACTION: - *Sherry Taylor*

Breakout Sessions
Group Two Summary:

We tried to distill it down at the end, to a couple of key take-aways as far as what we could talk through. The first is the enormity of what this project could be because there are so many stakeholder groups that could be involved and there is so much depth and breadth and variety of issues that affect all of these stakeholders. The more that we were talking the larger the issues became. But when we tried to come back as a group at the end of our time, we identified a couple of key take-aways.

One extraordinary opportunity that this group has is the idea of breaking some of those potentially negative themes/connotations around genetics and its implications and really challenge the idea that it is different than some other elements of the medical practice. How do we reframe genetics to the consumer away from the negative.

The second was building on the lessons learned. Because we have the history behind neonatal genetics, and in that process we have learned a variety of very important lessons. Where we are going in the future really builds upon that. We can use that as a guidepost, and we can build some case studies that reflect what we have learned, it really doesn't encompass where we are going because the disease categories are much larger and more complex. While they give us a history to build on, they might not always give us what we need to navigate the future.

Developing Case studies:

We built on the idea of both an antidepressant and possibly statins or gleevec, for a pharmacogenetics type of example.

We spent some time talking about the near-term/ far-term in the context of cancer because it's a diagnosed disease and thinking about some of the applications in genetics with a disease like that because the disease already exists, as opposed to some of the risk assessment type of applications where you are doing more prognostic type of work.

So the idea of potentially doing two different case studies, one in something that is more of a "here and now", such as a cancer example, and then something a little bit longer term, such as diabetes.

Definition of public health and the role of public health. The role the state potentially plays in initiatives like this and things that stakeholder groups would be interested in. And the role the state possibly plays in assuring that there are frameworks in place to get people up to speed on what they need to know.

We talked a little bit about revisiting the continuum of care and how since genetic information is going to be with you when you visit your primary care physician, if you happen to go to the ED, you need in-patient care, and making sure that whatever this turns into over time, there is a continuum of care. That the way we build lab testing modules for test reporting is applicable to that continuum of care.

In the context of the laboratory we talked about the “must have” at some point of some ability to critically evaluate specificity of sensitivity parameters around tests that are coming up and when they are applicable in certain care paths to begin to frame some protocols as far as how they could be used.

Recognizing that there may not be enough genetic counselors to be able to satisfy all of the demand moving forward, nor enough competently trained laboratorians, how do we empower, either with IT tools, or other type of care decision matrix tools that the general provider audience can use to do a better job of being a deliverer of genetic information?

What type of education tools can be developed that we, as a team could develop, that we could then take back to our core constituents that they could then use. It was almost a teach-the-teacher, we didn't reach an exact resolution as far as what those exact tools would be other than the case studies would have extreme value across the board because each can potentially resonate and hit a chord at a slightly different level of those core groups.

Group Two Notes: Case studies:
Antidepressants
Statins
Gleevac
Walk-through system, look for points of change for policy-makers to look at impact of genomics
Newborn screening example – also consider issue of screening for adult onset disease, Iding carriers (ie CF)
Look for lessons learned

Information needed:
What is the most effective testing? - protocols - sensitivity
What is the role of public health - agency - generally
Professional education – when, how
What makes a provider qualified? What continuing education is needed?
Education about what is an appropriate referral?
Importance of coordinated care after testing – who provides?
How are services being delivered?
Where do consumers want counseling provided? By whom?
What do consumers know/want to know about genetics?
IT tools to help providers
Barriers – stigma, fear of discrimination, perceived complexity – are they unique to genetics?
Is genetics different from general medicine?
Target different audiences with different educational approaches
Map out where genetic services are available
What restrictions are there on where genetic counselors can practice (ie licensing in California and Utah)
Diversity, cultural, language issues
Providers want cost effectiveness information, other facts.

Delivery of information:
Sent out to select members of organizations (beyond advisory group) for review.

Consumers:
Where in their state can they get information?
How will it impact the family unit?

ACTION: Distribute notes.
-Sherry Taylor

APPENDIX E

**Genetic Services Policy Project
Spring 2005 Advisory Meeting
Four Points Hotel Chicago O'Hare—April 8, 2005
Meeting Agenda**

10:00 a.m.

Introductions, Update & Discussion of Project Work

Rick Carlson, Grace Wang, Amber Roche, & Cindy Watts

11:00 a.m.

Genetics on the Radar Screen: Stakeholder Perspectives - Group and Individual Discussions

Rick Carlson

12:30 p.m.

—Lunch—

1:00 p.m.

Genetics on the Radar Screen: Stakeholder Perspectives - continuation

2:00 p.m.

—Break—

2:15 p.m.

Integrating Genetics into Consumer Information Products

Don Kemper, Healthwise

3:00 p.m.

Strategic Communication of Complex Social Issues

Frank Gilliam, UCLA

3:45 p.m.

Wrap-up

Rick Carlson

—Adjourn—

Summary Notes
Genetic Services Policy Project
Spring 2005 Advisory Committee Meeting
April 8, 2005
10:00 a.m. to 4:00 p.m.
Four Points Hotel Chicago O'Hare

The second advisory meeting for the Genetic Services Policy Project (GSPP) was held on Friday April 8th, 2005 at the Four Points Hotel Chicago O'Hare.

Attendees

Advisors/Consultants:

- Jean Anderson, International Society of Nursing Genetics
- Jeffrey Bauer, ACS Healthcare Solutions
- Meg Booth, Association of Maternal and Child Health Programs
- Amy Brin, American Academy of Pediatrics
- Daragh Conrad, National Society of Genetic Counselors
- Frank Gilliam, University of California Los Angeles
- Alissa Johnson, National Conference of State Legislatures
- Celia Kaye, Newborn Screening and Genetic Resource Center, University of Texas Health Science Center at San Antonio
- Don Kemper, Healthwise
- Ron Lankford, Healthcare Consultant to Payers
- Frances Margolin, Health Research and Educational Trust
- Joseph McInerney, National Coalition for Health Professional Education in Genetics
- Lawrence O'Connor, American Medical Association
- Inger Saphire-Bernstein, Blue Cross Blue Shield
- Peggy Stupca, Association of Genetic Technologists
- Leslie Wainwright, SG-2
- David Weismiller, American Academy of Family Physicians
- Mark Yarborough, University Colorado Health Sciences Center Center for Bioethics and Humanities

Project Investigators/Staff:

- Rick Carlson, University of Washington
- Penny Kyler, Department of Health and Human Services, Health Resources and Services Administration
- Amber Roche, Washington State Department of Health, Genetic Services Section
- Sherry Taylor, Washington State Department of Health, Genetic Services Section
- Grace Wang, University of Washington
- Cindy Watts, University of Washington
- Candi Wines, University of Washington

Not in attendance:

Advisors/Consultants:

- Bill Benedict, The Centers for Medicare and Medicaid Services, Region X
- Don Black, Child Health Corporation of America
- Carmella Bocchino, America's Health Insurance Plan
- Joanne Boughman, American Society of Human Genetics
- Brett Davis, IBM
- Patricia Deverka, Duke University
- Sarah Donta, The Council of State Governments
- Norman Kahn, American Academy of Family Physicians
- Trudi Matthews, The Council of State Governments
- Lee Newcomer, Ingenix
- Dan Perry, Alliance for Aging Research
- R.J. Ruff, The Centers for Medicare and Medicaid Services, Region X
- Priscilla Short, University of Chicago
- Barry Steinhardt, American Civil Liberties Union
- Sharon Terry, Genetic Alliance
- Michael Watson, American College of Medical Genetics

Project Investigators/Staff:

- Wylie Burke, University of Washington
- Kay Collins, University of Washington
- Debra Lochner Doyle, Washington State Department of Health
- Patricia Kuszler, University of Washington
- Michele Puryear, Department of Health and Human Services, Health Resources and Services Administration
- Scott Ramsey, University of Washington
- David Veenstra, University of Washington

The following represents a brief overview of the meeting and is not a detailed and complete transcript.

I. Introductions, Update, and Discussion of Project Work

Cindy Watts, PhD, Principal Investigator, Genetic Services Policy Project and Rick Carlson, JD, Investigator, Genetic Services Policy Project welcomed the Advisory Committee members and gave a brief overview of the goals for the three year Genetic Services Policy Project (GSPP). Year one will be complete in May 2005; the goals for this year were to collect information and begin to describe available genetic services.

The first product from this year's efforts is a cost effectiveness analysis literature review titled, "A Systematic Review of Economic Evaluations of Genetic Testing Technologies". GSPP investigators will present the literature review at two conferences: 1) International Society for Pharmacoeconomics and Outcomes Research, Washington, DC, May 2005; and 2) International Health Economics Association, Barcelona, Spain, July 2005. Genetics in Medicine has also accepted the literature review for publication.

The second product from Year 1 is a draft of eight state profiles describing genetic services by state. Staff chose seven of the states to represent the seven regional collaborations; Washington was included as the eighth state because of the staff's familiarity with services in Washington State. The Advisory Committee received the draft document in order to provide comments. Copies were included in the meeting binders.

The goals of years two and three include completing case studies and disseminating information to trade and professional organizations. The GSPP will invite the Advisory Committee: 1) To participate in key informant interviews; 2) To provide guidance in the development of products for dissemination; and 3) To participate in workshops to present information.

Materials developed by GSPP are available for use by Advisory Committee members upon request to project staff.

Genetic Services Delivery

Amber Roche, MPH, Washington State Department of Health, Genetic Services Section

Ms. Roche presented data on the delivery of genetic services. Handouts of her slide presentation were included in the meeting binders. The discussion following the presentation included comments on the need to integrate genetics training in medical school curricula. Genetics is relevant to many specialties and should be taught as an integrated topic. The observation was made that medical doctors tend to choose other, more lucrative specialties over genetics. A suggestion was made to compare the number of specialists per capita that practice genetics to the number of other medical specialists per capita such as pediatricians, cardiologists, etc. It was noted that data on who receives genetic services are difficult to obtain. For example, databases using ICD-9 and CPT codes are not very informative because genetic services might not have unique codes. Instead, they may be lumped into general categories or codes as 'other' services. Results from the Genetic Alliance survey may be a good source of data about who receives services.

Genetic Activities in Public Programs: A Baseline Inventory

Grace Wang, MPH, University of Washington

Ms. Wang presented a summary of findings based on an inventory of genetic services provided or sponsored by state agencies. Her presentation included information on the types of services provided, how they are delivered, and who pays for them. Handouts of her slide presentation were included in the meeting binders.

Suggestions were made on where to obtain relevant data. For example, Maternal and Child Health block grant recipients report the incidence of specific genetic disorders detected through newborn screening programs; therefore each state program should have this information.

The group discussed the concept that everything has a genetic component. It was suggested that an appropriate question might be, “what is the role of genetics in a specific disease or in the spectrum of a disease?” The variability in gene expression, the number of genes that might influence a single disease, and gene-environment interaction affect the degree to which genes influence a disease process. It was noted that as more is revealed about the human genome, we are moving from identifying single gene disorders to trying to understand common diseases with complex genetic components. The group discussed the risks of ‘overselling’ the role of genetics and genetic exceptionalism.

The discussion also included comments about consumer perception and how it has the potential to drive the field. Consumer demand, or lack of it, can dictate the development and utilization of genetic tests and services. If consumers believe that ‘genetics’ is the cause of their conditions, will they be willing to make behavior or environmental changes or will they expect a medical fix?

Other business:

Alissa Johnson made two announcements: 1) NCSL is developing a booklet for state legislatures on states’ roles in genetic programs and 2) NCSL is having its annual conference in Seattle on August 16-20, 2005 and is considering having an MCH roundtable discussion. Contact Alissa Johnson (alissa.johnson@ncsl.org) for more information.

II. Genetics on the Radar Screen: Stakeholder Perspectives – Group Discussion

The discussion continued with a question about whether payers emphasize or would expect to emphasize newborn screening. The group also considered actual risks for genetic discrimination versus perceived risks, and whether actual or perceived risks affect medical record-keeping.

Members of the committee noted that patients and providers are reluctant to report having a genetic test or the result of a genetic test. In addition, the lack of accurate ICD-9 and other billing codes limits the specificity of available information. This presents challenges for insurance companies, which use information about service utilization to develop benefit plans and set rates. Insurance companies often use proxy data if actual data are not available (e.g. family history).

The group noted a need for federal legislation to limit employment and insurance discrimination. Various drafts of legislation have been proposed at the federal level for the past several years; none of the bills have passed both houses.

In the future, genetic services may be used more for conditions such as cancer and rheumatoid arthritis to determine gene expression and tumor types rather than to detect and diagnose heritable conditions. This suggests that more patients will rely on specialists in other fields (e.g. oncology, rheumatology) than on geneticists. It also portends a need for a spectrum of genetic services over the course of a disease (e.g. screening, diagnosis, treatment, and monitoring).

The existing education system does not have the capacity to train health care providers for this type of care. There is a need for training that cross-cuts disciplines and relies on shared resources and joint curricula. The current education model is also influenced by licensing exam structures and requirements; there is an emphasis on “being an expert.” It was noted that both integrated and siloed training programs may be necessary; there will continue to be a need for experts and specialists.

Members shared some personal experiences with using integrated training models. Schools that advertise multidisciplinary programs have been shown to be more successful at recruiting students than schools that do not

offer multidisciplinary programs. A program that trains laboratory technicians to work in multiple specialties is very successful. On the other hand, an effort to integrate genetics into basic nursing training curricula has been only minimally successful. Other countries may have education and training models that are better at producing cross-trained professionals.

III. Integrating Genetics into Consumer Information Products

Information Therapy in the World of Genomics

Donald W. Kemper, Chairman and CEO, Healthwise

Mr. Kemper led a discussion on how consumer information products are developed and how their use is promoted by physicians, payers, and consumer groups. Copies of his slides were distributed at the meeting. He described the importance of “information therapy” (Ix) as a component of a patient’s care. Information therapy is the method of delivering the right information at the right time to help someone make the right decision. A critical element of the success of information therapy is having an electronic medical record.

One member asked how patients will manage risk information versus diagnostic information and be able to keep it private, yet share it with providers to receive the appropriate care. Testing for BRCA1/2 mutations was used to illustrate the impact of risk information on patient decision-making. A member noted a lack of both legal and financial support to treat risk rather than disease.

Payers were asked if they were developing policies that determine when and if they will cover certain procedures. Blue Cross/ Blue Shield plans use an evidence-based model to determine which services they cover. The procedure must show clinical validity and clinical utility.

Many clinical geneticists practice information therapy, but they are not compensated for the time and effort.

The risk of medicalizing everything or making genetics a part of everything and the possibility of losing the ability to change personal behaviors was discussed again in the context of this model. Some committee members noted that there are inadequate tools to study this phenomenon, especially in relation to genetics where the route from genotype to phenotype is not always well understood.

There were questions about how effective the information therapy model is and if the effectiveness depends on the education level of the patient. In general, studies are showing that this type of approach is effective. Studies of other interventions have shown that people will use information if it is offered, easy to get, and appropriately designed.

One of the biggest expenses involved in educating consumers is direct to consumer (DTC) marketing. An important factor in the success of DTC is whether a physician supports a product and its advertised message. The development, supply, and utilization of genetic technologies may be a “bottom-up movement” driven by consumer demand. Increases in discretionary spending combined with DTC marketing and the proliferation and movement of information are all factors that could contribute to a consumer-driven market for genetic services.

Something to consider: Are there circumstances under which a consumer should be prohibited from having information about his/her health?

IV. Strategic Communication of Complex Social Issues

Communicating Complex Social Issues: The Public Discourse about Early Child Development

Dr. Franklin D. Gilliam Jr., University of California, Los Angeles

Dr. Gilliam gave a presentation on how to construct messages and information to fit within a frame through which the public can relate and understand the message. He emphasized how to talk about the issue rather than focusing on the audience. Copies of his slides were included in the meeting binders.

Dr. Gilliam used an example of framing information about early child development. Other examples where this model has been successful include a children's oral health campaign, constructing messages about environmental health, and framing argument during the tobacco law suits.

The media play a substantial role in framing issues. It is difficult to combat the media's messages without the financial resources they have. One approach is to re-orient journalists, find out who their sources are and give them other sources that can deliver a different message.

V. Wrap Up

Dr. Watts solicited ideas for the next meeting. They included:

- 1) Bring a commercial testing perspective
- 2) Look broadly at educational issues
- 3) Invite ACMG and genetic counselor views on the future of genetic services, pressures of delivering genetic services, and how the genetic system needs to adapt.
- 4) Provide information about how tests are validated
- 5) Bring information or have a discussion about whether liability or malpractice change with the availability of more tests and information
- 6) Have a discussion about why public health may not want or be able to move beyond newborn screening
- 7) Discuss the licensure of genetic counselors and how it might change service delivery
- 8) Present information from the HRSA Personal Health Tools (PeHT) project on how the Internet affects health care
- 9) Discuss the role of the religious right in genetic issues
- 10) David Weismiller offered to give an update about the AAFP genetics year—education and uptake
- 11) Amy Brin offered to give an update on co-management