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## Genetic Services Policy Project Final Report

### Appendix A: Dissemination Activities

Dissemination of results has been a major component of the **Genetic Services Policy Project**. The dissemination activities have taken many forms, from peer-reviewed publications and workshops to informal articles and presentations. The GSPP has attained a high rate of acceptance for peer-reviewed submissions—85 percent, or 34 out of 40. Project personnel have also taken part in 15 informal presentations and six written dissemination activities. Finally, we have held four in-person meetings of the national advisory committee, produced quarterly newsletter updates, and provided website resources. A summary count follows; see Appendix A.1 (page 3) for a full list.

#### Genetic Services Policy Project Dissemination Activities

- 8 published articles (peer-reviewed)
- 3 published articles (not peer-reviewed)
- 12 poster presentations (peer-reviewed)
- 14 formal presentations, including 7 workshops
- 15 informal presentations
- 6 informal written dissemination activities
- 6 peer-reviewed submissions that were not accepted
- 13 formal communications with advisory committee, including 4 annual face-to-face meetings

GSPP has produced eight peer-reviewed publications in the following journals: *American Journal of Managed Care*, *American Journal of Public Health*, *Genetics in Medicine*, *Health Affairs*, *Journal of Health Politics, Policy and Law*, and *Maternal and Child Health Journal*. Articles include topics such as economic analyses and evidence-based assessments of genetic services, genetics policy, utilization trends of genetics clinics, and genetic counseling for children with special health care needs. The 12 peer-reviewed posters address similar subjects. A list of article titles and authors is included in Appendix A.1.

GSPP has given a number of formal presentations. The conference organizers include American College of Medical Genetics, American Public Health Association, Centers for Disease Control and Prevention, Genetic Alliance, National Institutes of Health, and World Research Group. Dates and titles of presentations are included in Appendix A.1.

With the assistance of many members of our Advisory Committee, we developed a series of workshops that were delivered at meetings and conferences of some of the key trade associations represented by our advisors: AdvaMed, America's Health Insurance Plans, American Hospital Association, American Medical Association, Association of Genetic Technologists, Biotechnology Industry Organization, and Child Health Corporation of America. Between November 2006 and October 2007, we delivered seven workshops. The workshops allowed us to directly disseminate our work to hundreds of providers, managers and executives

in the health care delivery system. The response to our material was excellent, and those who attended felt that this was a very efficient method of dissemination. For those workshops that conducted evaluations, GSPP's presentations received consistently positive feedback. The presentation and evaluation materials from each workshop appear in Appendix A.2 (page 9).

Project personnel have also participated in a number of informal presentations to audiences such as the Western States Genetic Services Collaborative, the National Coordinating Center for the Regional Genetics and Newborn Screening Collaborative Groups, Washington State Genetics Providers Group, Washington State Perinatal Advisory Committee, the Institute of Medicine Cancer Policy Forum, and units within the University of Washington, the National Human Genome Research Institute, and the National Society of Genetic Counselors.

GSPP has taken advantage of informal opportunities to disseminate written materials to the National Conference of State Legislatures, the National Newborn Screening and Genetics Resource Center, and several chief medical officers. We sent a letter to the Secretary's Advisory Committee on Genetics, Health and Society, responding during the public comment period to the draft report, "Policy Issues Associated with Undertaking a Large U.S. Population Cohort Project on Genes, Environment, and Disease." We also had an informational display table at the National Society of Genetic Counselors' Annual Education Meeting. In addition, our website showcases GSPP's project updates and resources.

In sum, our dissemination activities have reached a broad audience on a range of topics related to genetic services. Our materials have been well received, as evidenced by our high publication success rate and the uniformly positive evaluation of our presentations.

## Appendix A.1: Complete List of Dissemination Activities

### ***Papers and Articles (peer-reviewed)***

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
October 2005	Article	Economic analyses of human genetics services: a systematic review	<i>Genetics in Medicine</i> . 2005 Oct;7(8):519-523.	Josh Carlson
October 2005	Article	The Case of BiDiI: A Policy Commentary on Race and Genetics	<i>Health Affairs</i> , web exclusive, October 11, 2005	Rick Carlson
April 2006	Article	Toward Evidence-based Assessment for Coverage and Reimbursement of Laboratory-based Diagnostic and Genetic Tests	<i>Am J Manag Care</i> . 2006;12:197-202	Rick Carlson, David Veenstra
February 2007	Article	Genetic Counseling, Insurance Status, and Elements of Medical Home: Analysis of the National Survey of Children with Special Health Care Needs	<i>Maternal and Child Health Journal</i>	Grace Wang
April 2007	Article	The role of genetics in the provision of essential public health services	<i>American Journal of Public Health</i> 97(4):620	Grace Wang
October 2007	Article	Utilization trends of genetics clinics excluding prenatal services in Washington State, 1995 to 2004	<i>Genetics in Medicine</i> . 9(10):713-718.	Grace Wang, Cindy Watts
October 2007	Article	Pharmacogenomic testing to prevent aminoglycoside-induced hearing loss in cystic fibrosis patients: potential impact on clinical, patient, and economic outcomes	<i>Genetics in Medicine</i> . 9(10):695-704.	Dave Veenstra
February 2008	Article	Preemptive Public Policy for Genomics	<i>Journal of Health Politics, Policy and Law</i> 33: 39-51	Rick Carlson

### ***Papers and Articles (not peer-reviewed)***

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
July 2004	Article	Describing GSPP	National Society of Genetic Counselors' newsletter <i>Perspectives</i>	Deb Lochner Doyle Amber Roche
May 2005	Article	Describing GSPP	Association of Maternal and Child Health Programs newsletter <i>Pulse</i>	GSPP personnel
June 2006	Article	Third Party Payer Perspectives	Health Industry Forum Foundation	Rick Carlson

**Posters (peer-reviewed)**

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
May 2005	Poster presentation	A Systematic Review of Economic Evaluations of Genetic Testing Technologies	International Society for Pharmacoeconomics and Outcomes Research in Washington, D.C.	Scott Ramsey
July 2005	Poster presentation	A Systematic Review of Economic Evaluations of Genetic Testing Technologies	International Health Economics Association in Barcelona, Spain	Scott Ramsey
October 2005	Poster presentation	Organization, Administration and Funding of State Genetic Services Programs	American Society of Human Genetics Annual Meeting in Salt Lake City, UT	Grace Wang
March 2006	Poster presentation	Pharmacogenomics and aminoglycoside-induced hearing loss in cystic fibrosis patients	American College of Medical Genetics Annual Clinical Genetics Meeting in San Diego, CA	Julie Harris
September 2006	Poster presentation	Utilizations of Genetics Clinics in Washington, 1995 – 2004	University of Washington Annual Gathering of Health Sciences Schools incoming students	Grace Wang
November 2006	Poster presentation	Factors associated with need and use of genetic counseling: An analysis of the National Survey of Children with Special Health Care Needs	American Public Health Association Annual Meeting in Boston, MA	Grace Wang
November 2006	Poster presentation	Policy implications of cost effectiveness analysis for genetic testing: A case study	American Public Health Association Annual Meeting in Boston, MA	Julie Harris
November 2006	Poster presentation	Integrating Personalized Medicine into the Health Care System: A Comprehensive Study of Stakeholder Groups	American Medical Association meeting	Patricia Deverka (GSPP Advisor)
November 2006	Poster presentation	Through the media lens: Analyzing the news messages Americans receive about genetics	National Society of Genetic Counselors' Annual Education Conference in Nashville, TN	Amber Roche
November 2006	Poster presentation	Integrating genomics into the healthcare system: opportunities for collaborations by stakeholders	National Society of Genetic Counselors' Annual Education Conference in Nashville, TN	Grace Wang Amber Roche
November 2006	Poster presentation	Proving Genomics: A Workshop for Physicians	Accepted – Society of Teachers of Family Medicine Annual Spring Conference in April in Chicago	Rick Carlson
March 2007	Poster presentation	Utilization Trends of Genetics Clinics in Washington State, 1995 to 2004	American College of Medical Genetics annual meeting in Nashville, TN, in March 2007	Grace Wang

### **Formal Presentations and Workshops**

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
September 2005	3 Panel presentations	Payment and reimbursement	Access to Quality Testing for Rare Disorders: National Institutes of Health (NIH) National Conference in Rockville, MD	Rick Carlson
March 2006	Presentation	Evaluation of Genomic Applications in Practice and Prevention (EGAPP) assessment of genetic testing; program review and follow-up	Centers for Disease Control and Prevention in Atlanta, GA	Rick Carlson
March 2006	Presentation	Stakeholders' Perspectives on the Delivery of Genetic Services	American College of Medical Genetics Annual Clinical Genetics Meeting in San Diego, CA	Rick Carlson, Deb Lochner Doyle
July 2006	Presentation	Personalized Medicine Landscape: challenges and opportunities for consumers	Genetic Alliance Annual Conference in Bethesda, MD	Rick Carlson
October 2006	Presentation	Quality, Access, and Sustainability of Biochemical Genetic Testing	Joint National Institutes of Health (NIH) Office of Rare Diseases – Centers for Disease Control and Prevention in Atlanta, GA	Rick Carlson, Michele Puryear
November 2006	Presentation	A model for determining the budget impact of expanded newborn screening	American Public Health Association Annual Meeting in Boston, MA	Wylie Burke
November 2006	Presentation	Genomics Stakeholder Workshop	American Medical Association meeting	Rick Carlson, David Veenstra
November 2006	Presentation	Genomics Stakeholder Workshop	AdvaMed meeting – Molecular Diagnostics: Development, Regulation, & Reimbursement	Rick Carlson, Jeff Bauer (GSPP advisor)
April 2007	Presentation	Transforming Health Care: The emergence of Genetic Services in Medicine and Health Care	Child Health Corporation of America	Rick Carlson
May 2007	Presentation	Proving Genomics: What Do Payers and Providers Need?	BIO (Biotechnology Industry Organization) International Convention, Boston, MA	Rick Carlson, Jeff Bauer (advisor)
June 2007	Presentation	Proving Genomics	Association of Genetic Technologists' Annual Meeting, Denver, CO	Rick Carlson, Deb Lochner Doyle
June 2007	Presentation	Proving Genomics: A Workshop for Payers	America's Health Insurance Plans Annual Meeting in Las Vegas, NV	Rick Carlson
July 2007	Presentation	Integrating Genomics: What Do Hospitals Need?	American Hospital Association / Health Forum's Leadership Summit in San Diego, CA	Cindy Watts
November 2007	Presentation	Integrating Medical Genetics and Genetic Testing to Enhance the Value of Healthcare Delivery and Disease Prevention	Genetic Testing & Genetic Risk Assessment for Health Plans, World Research Group	Rick Carlson

### ***Informal Presentations and Workshops***

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
May 2005	Presentation	Year 3 proposal narrative as background for a task force	National Society of Genetic Counselors' Genetic Services Delivery Task Force	Daragh Conrad (GSPP Advisor)
September 2005	Web cast presentation	Financing of Genetic Services based on GSPP State Genetics Profiles data	National Coordinating Center for the Regional Genetics and Newborn Screening Collaborative Groups	Deb Lochner Doyle
September 2005	Conference participation	Genetics and newborn screening services	National Coordinating Center for Regional Genetics and Newborn Screening Collaborative Groups in Washington, D.C.	Grace Wang
September 2005	Presentation	Project update	Western States Regional Genetics Collaborative meeting in Las Vegas, NV	Amber Roche
December 2005	Discussion / presentation	Integrating genetic services into current system	Joint Centers for Disease Control and Prevention-Group Health Cooperative meeting in Seattle, WA	Rick Carlson
February 2006	Presentation	Assessed databases, described GSPP activities	GeneTests in Seattle, WA	Rick Carlson
March 2006	Presentation	GSPP state genetics profiles, case studies, and cross-cutting policy format	Washington State Genetics Provider Group in Kent, WA	Deb Lochner Doyle
June 2006	Presentation	Project update	HRSA offices in Washington, D.C.	Rick Carlson
June 2006	Discussion	Reimbursement policy	National Human Genome Research Institute	Rick Carlson
June 2006	Utility, infrastructure	Developing guidance process for evidence-based coverage decisions regarding genetic services	Diagnostic developers and payers	Rick Carlson
June 2006	Briefing attendance	A Bill to improve and expand the use of molecular genetic tests and therapeutics	Senator Barack Obama's staff in Washington, D.C.	Rick Carlson
October 2006	Presentation	Update on the work of the Genetic Services Policy Project	Western States Genetic Services Regional Collaborative Summit	Amber Roche
October 2006	Presentation	Preemptive Public Policy for Genomics	UW Dept of Health Services Policy Seminar Series	Rick Carlson
February 2007	Presentation	Genetic Services Policy Project findings	Washington State Perinatal Advisory Committee	Deb Lochner Doyle
March 2007	Presentation	Billing and reimbursement issues for genetic services	Institute of Medicine Cancer Policy Forum	Deb Lochner Doyle

### **Informal Written Dissemination**

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
August 2005	Fact sheets	GSPP overview	National Conference of State Legislatures Annual Meeting in Seattle, WA	Candi Wines
September 2005	Fact sheets	Genetic Services Policy Project overview	Requested by and sent to 7 advisors and chief medical officers	Candi Wines
September 2005	Template – state genetics profiles	State genetics profiles and GSPP overview	Sent to National Newborn Screening and Genetics Resource Center	Candi Wines
October 2005	Flow chart diagrams	Organization, Administration and Funding of State Genetic Services Programs	Sent to National Conference of State Legislatures for a booklet they are creating	Grace Wang
July 2006	Public Draft Comment Letter	Comments on public draft of SACGHS' report: Policy Issues Associated with Undertaking a Large U.S. Population Cohort Project on Genes, Environment, and Disease	Secretary's Advisory Committee on Genetics, Health, and Society	GSPP personnel
November 2006	Display table	GSPP State Genetic Profiles, Fact Sheets, Informational Tear-away Sheets	National Society of Genetic Counselors' Annual Education Conference in Nashville, TN	Amber Roche

### **Submissions That Were Not Accepted**

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
June 2005	Panel presentation	Impact of genetic services on disparities in vulnerable populations	Submitted to Grantmakers in Health	Rick Carlson
October 2005	Panel presentation	The role of genetic services in health care: barriers and opportunities	Submitted to American Society of Human Genetics Annual Meeting	Deb Lochner Doyle
October 2005	Article	Expenditures for Genetic Services in the United States: An Overview	Submitted to <i>Health Affairs</i>	Grace Wang
January 2006	Article	Stakeholders' perspectives regarding genetics and genetic services in the United States	Submitted to <i>Clinical Genetics</i>	Grace Wang
January 2006	Panel presentation	Policy Research on Genetic Services: New Science Meets Old Paradigm	Submitted to Academy Health Annual Research Meeting	Carolyn Watts, Deb Lochner Doyle
April 2006	Poster	Promoting Health by Integrating Genomics into the Healthcare System: Opportunities for Collaborations by Stakeholders	Centers for Disease Control and Prevention annual National Health Promotion Conference	Grace Wang
Continuous	Website	GSPP updates and resources	GSPP's website	GSPP personnel

### **Dissemination to the Advisory Committee**

<b>Date</b>	<b>Activity</b>	<b>Topic</b>	<b>Venue</b>	<b>Primary Person</b>
September 2004	Presentations / Conference	Update to Advisory Committee on GSPP Year 1	Annual Advisory Committee Meeting, Washington D.C.	Project personnel
April 2005	Presentations / Conference	Update to Advisory Committee on GSPP Year 1	Annual Advisory Committee Meeting, Chicago, IL	Project personnel
June 2005	GSPP update	Annual summary sent to advisors	Via email	GSPP personnel
September 2005	GSPP update	Quarterly communication with advisors	Via email	GSPP personnel
November 2005	GSPP update	Quarterly communication with advisors	Via email	GSPP personnel
April 2006	Presentations / Conference	Update to Advisory Committee on GSPP Year 2	Annual Advisory Committee Meeting, Washington D.C.	Project personnel
June 2006	GSPP update	Annual summary sent to advisors	Via email	GSPP personnel
August 2006	GSPP update	Quarterly communication with advisors	Via email	GSPP personnel
February 2007	GSPP update	Quarterly communication with advisors	Via email	GSPP personnel
April 2007	GSPP update and review	Quarterly communication with advisors	Via email	GSPP personnel
May 2007	Presentations / Conference	Update to Advisory Committee on GSPP Year 3	Annual Advisory Committee Meeting, Washington D.C.	GSPP Personnel
August 2007	Backgrounder distribution	Backgrounder on Pharmacogenomics for the Pharmaceutical and Biotechnology Industries: Basic Science, Future Scenarios, Policy Directions	Mailed a copy of backgrounder to each GSPP advisor	Rick Carlson
October 2007	GSPP update	Quarterly communication with advisors	Via email	GSPP personnel

**Appendix A.2: Summary of Evaluations from Genomics Workshops**

<b>Organization</b>	<b>Conference Title</b>	<b>Location, Date</b>	<b>Workshop Title</b>	<b>Presenters</b>
American Medical Association	2006 Interim Meeting of the AMA House of Delegates	Las Vegas, NV November 12, 2006	Proving Genomics: A Workshop for Physicians	Rick J. Carlson, JD, Genetic Services Policy Project  David Veenstra, Pharm D., PhD, Genetic Services Policy Project
AdvaMed	Molecular Diagnostics: Development, Regulation, and Reimbursement	Washington, DC November 16, 2006	Proving Genomics: A Stakeholder Workshop	Rick J. Carlson, JD, Genetic Services Policy Project  Jeffrey C. Bauer, PhD, Senior Vice President, ACS Healthcare Solutions  Paul Campbell, Managing Director, EBG Advisors
Child Health Corporation of America	Strategic Planning and Business Development Forum	Los Angeles, CA April 26, 2007	Transforming Health Care: The emergence of Genetic Services in Medicine and Health Care	Rick J. Carlson, JD, Genetic Services Policy Project
Biotechnology Industry Organization	BIO International Convention – New Ideas: Bold Ventures, Global Benefits	Boston, MA May 6-9, 2007	Proving Genomics: What do Payers and Providers Need?	Rick J. Carlson, JD, Genetic Services Policy Project  Jeffrey C. Bauer, PhD, Senior VP, ACS Healthcare Solutions  Paul M. Campbell, MS, Managing Director, EBG Advisors
Association of Genetic Technologists	32 <sup>nd</sup> Annual Meeting	Denver, CO May 31-June 3, 2007	Integrating Genetic Services into Mainstream Health Care	Rick J. Carlson, JD, Genetic Services Policy Project  Debra Lochner Doyle, MS, CGC, Genetic Services Policy Project
America's Health Insurance Plans	Annual Meeting	Las Vegas, NV June 20-22, 2007	Proving Genomics: A Workshop for Payers	Rick J. Carlson, JD, Genetic Services Policy Project  Allan J. Ebbin, MD, MPH, Vice President, Healthcare Quality and Education, Sierra Health Services
American Hospital Association	2007 AHA/Health Forum Leadership Summit	San Diego, CA July 22-24, 2007	Integrating Genomics: What Do Hospitals Need?	Carolyn Watts, PhD, Genetic Services Policy Project  Marc Williams, MD, Director, Clinical Genetics Institute, Intermountain Healthcare

***American Medical Association Evaluation***

Audience: 70 senior level physicians

Evaluation: The AMA used its own evaluation form following the presentation.

	Strongly Agree			Strongly Disagree	
	1	2	3	4	5
The program was well organized.	31	23	3	2	1
The learning objectives were adequately addressed.	25	26	4	4	0
I can now discuss the evolving technology of genomics.	12	31	14	3	0
I can now describe the current scope of genetic testing services.	13	31	13	3	0
I can now discuss the findings and policy recommendations of the Genetic Services Policy Project.	8	25	18	5	2
I can now describe the clinical utility of personalized medicine.	13	32	13	2	0
I can now identify the uses of genetic data for predictive testing.	10	31	14	5	0
I can now describe the fundamentals of pharmacogenomics.	15	30	12	3	0
	Yes			No	
Did you perceive any commercial bias in the presentation?	1 (No explanation given)			54	

**Comments/Suggestions:**

Several “outstandings.” “Excellent, exciting program.” “More of the same.” “Update annually.” “Excellent presentations by Rick Carlson and David Veenstra.” “Very Good, thanks!” “Very interesting – Good to get real scoop on what is/isn’t out there and how rapidly (or not) this is likely to unfold.” “No current impact.” “Great speakers, excellent information, well done!” “Tough topic, very complex.” “Great discussion.” “Liked combination of speakers and having socio-economic context for the technical information.” “Good overview – not much news.” “I have a real interest in slow metabolizers and that was very helpful.” “Wish more physicians attended and participated. This is an important field. Thanks for the good handouts.”

“Expand discussion of prediction testing. How can technology help people alter their lifestyle?” “Less quotations and predictions, more about present facts and uses.” “More details.” “Mention proteomics.” “Provide list of references.” “Explore more generalized genetic testing (mitochondrial DNA) in exploring racial/genetic background of individuals. “Time lapse prediction” “Longer, more depth” “Information that is currently not available, more specifics re: testing and uses.”

TOTAL OF 64 CME forms returned.

### *AdvaMed Evaluation*

Audience: The audience numbered about 200, and was comprised of senior managers, mostly marketing, product and business development from various diagnostic companies.

Evaluation: AdvaMed did not conduct presentation evaluations.

### *Child Health Corporation of America Evaluation*

Audience: 30 strategic planning and business development employees

Evaluation: CHCA collected written comments and feedback from attendees:

- Engaging - intriguing - thought provoking – GREAT
- Thank You Thank You Thank You - Brilliant, Helpful, Fabulous
- Very enlightening, Excellent, liked his approach. We need to keep this item on our agenda for future meetings.
- Would have liked more time.
- Very interesting!
- Unclear how message can be taken back for some sort of implementation.
- Wonderful view of the future.
- Awesome - though provoking.
- I'd love to hear more on this - maybe do some roundtables on it?
- This was a very informative and thought provoking lecture.

### *Biotechnology Industry Organization Evaluation*

Audience: There were approximately 300 biotechnology executives in attendance.

Evaluation: BIO did not conduct presentation evaluations.

### *Association of Genetic Technologists Evaluation*

Audience: There were approximately 20 genetic technologists in attendance.

Evaluation: AGT conducted its own presentation evaluations.

	Excellent 4	Good 3	Fair 2	Poor 1
Speaker Preparation	9	1	0	0
Level of Seminar	7	3	0	0
Length of Seminar	7	3	0	0
Effective Presentation of Materials	7	3	0	0
Usefulness of Handout Materials	4	5	0	0

#### **What is the most valuable insight gained from this seminar?**

- What we do as clinical medicine is a small part of the overall medical system.
- A look at genetics in overall healthcare system.
- The issues of current and future delivery. This was an excellent presentation.
- This should be repeated next year – presented to entire conference attendance in Houston.

- Public perspective, policy and pharmaceutical impact for PGX.
- Impact of personalized medicine.
- The whole lecture was very educational.
- So much information available.
- We need to educate more about genetics.

**Will you use information gained in this seminar in your professional position?**

Yes: 8                      No: 1

**What was your main motivation for attending this particular seminar?**

- To be educated on how genetics will eventually effect the medical system.
- Clinical background as well as cytogenetics and concerns about how the science reaches the public and how to educate providers.
- To learn more about genetics.
- Topic relevance.
- PGX status.
- Interested in the topic.
- Interest – coevolution of genetics with medicine.
- To be more educated about the field.

**Did this seminar meet or exceed your personal goals and expectations?**

Yes: 9                      No: 0

**Why or why not?**

- Very thought provoking.
- It was easy to follow along and full of lots of information.

***America’s Health Insurance Plans Evaluation***

Audience: There were approximately 150 managed care executives in attendance.

Evaluation: AHIP conducted a short evaluation:

Both speakers received the same scores (Good is a 3 on a scale of 1 to 4).

Speaker Effectiveness:    Good  
 Presentation Content:    Good  
 Value to Organization:    Good

***American Hospital Association Evaluation***

Audience: There were approximately 30 hospital leaders in attendance.

Evaluation: AHA conducted a short evaluation:

Excellent:                  2 responses  
 Very Good:                5 responses  
 Good:                        7 responses  
 Satisfactory:              1 response

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### **Appendix B: Analysis of GINA Through the Advocacy Coalition Framework**

The Genetic Information Nondiscrimination Act of 2007 (GINA) would have seemed to be legislation with no impediment to passage in the 110<sup>th</sup> Congress. Previous versions of GINA had passed the Senate by big margins in 2003 (108<sup>th</sup> Congress) and 2005 (109<sup>th</sup> Congress). President Bush had been favorable to passage and had indicated that he would sign the legislation into law.

The House of Representatives passed GINA in February 2007 by a wide margin. The House version of GINA, sponsored by Representative Louise Slaughter (D-NY), had been blocked in previous years by the Republican leadership that controlled the House. Democratic control of the House following the 2006 midterm Congressional election s cleared the way for house passage and – or so it seemed – quick passage in the Senate.

And yet as of the spring of 2008, GINA had not come to a vote in the Senate and there was no apparent prospect of a Senate vote on GINA in the near future. Indeed, there seemed a substantial likelihood that the 110<sup>th</sup> Congress would adjourn without the Senate having taken up this legislation that it had passed twice before by wide margins, and that – this time – the House *had* acted upon. When the 2007 version of GINA had been enacted by the House and sent on to the Senate for consideration it was subjected to a “hold” by Senator Tom Coburn (R-OK), and Coburn’s objections (based on objections to GINA raised by business interests and the U.S. Chamber of Congress) seemed to be irresolvable.

Then, in early April, 2008, there began to be indications that after a year with no visible activity, behind-the-scenes negotiation by a very persistent Senator Ted Kennedy (D-MA) – with the constant support of pro-GINA interests led by the Center for Responsible Genetics and the Genetic Alliance – was leading to a compromise that would move GINA to a vote in the Senate. On April 24<sup>th</sup> the Senate passed a modified version of GINA by a vote of 95-0. On May 1<sup>st</sup> the House passed the Senate version of GINA by a vote of 414-1, and the legislation was signed into law by President’s Bush on May 21, 2008.

Why GINA was floundering in the 110<sup>th</sup> Congress bears examination as a case study of national policy-making, a study of the manner in which interest groups affect policymaking, and a study of the agenda-setting that occurs at the federal policy-making level. In particular, GINA as a case study illustrates the value of Paul Sabatier’s Advocacy Coalition Framework as a shorthand method for understanding the policy process.

#### **An Introduction**

For citizens to have meaningful participation in the US political economy, they must find ways to act and speak collectively. Interest groups represent the freedom to join together with others to make political contributions and demands. These groups must impact the politicians and bureaucrats who serve as our policymakers, within the structure of our governing institutions.

Two concepts have come to describe our governing institutions and the intersecting activities of interest groups: political subsystems and issue networks.

The concept of the political subsystem is recognition that much of our policy-making takes place at levels below the three classical branches of American government, predominately at the administrative level. Surrounding the activities of this administrative state are issue networks, loose structures involving various bureaucrats, interested academics and professionals, and interest group representatives, all with a mutual interest in matters of policy-making within the subsystem.

As agenda-setting raises issues to the level of policy-making probability, these interested parties and interest group representatives form advocacy coalitions to stake claim to influence on the politics and policies at issue. Political and policy entrepreneurs also respond in such agenda-setting moments, to try and stake claims of influence on the policies at issue.

### **The Genetic Information Non-Discrimination Act of 2007**

There has been concern over issues of “genetic privacy” (protection against the misuse of genetic information by employers and health insurers) for over twenty-five years (Hudson, 2007). Some states enacted statutes in the 1970s that prohibited discrimination in health insurance underwriting based upon positive sickle cell and other genetic traits. Interest in these issues increased dramatically when Congress funded the commencement of the Human Genome Project in 1990. The first consideration of federal legislation – the 1990 Human Genome Privacy Act – was directly tied to the beginning of the Project. With the conception of the Human Genome Project, efforts were led by the Center for Responsible Genetics, based in Cambridge, Massachusetts, to get state legislation enacted that would prohibit the use of genetic information to deny healthy individuals insurance or employment. Broad legislation was passed in California, but vetoed by Governor Pete Wilson, who expressed concern for adverse effects on employers providing employee health insurance. Broad state legislation was adopted in Oregon, and went into effect in 1995, but even with regard to that legislation the potential for adverse employment and health insurance actions remained.

As scientific research continued throughout the 1990s leading to the completion (in 2001) of the mapping of the genome, debate continued over concerns that the ultimate predictive medical advances to be gained by the Human Genome Project would pale by comparison to the negative impacts of the use of genetic information by employers and health insurance underwriters. Some commentators pointed out that state legislation regarding health insurance underwriting would not apply to self-insured employers because of the preemption provisions of the federal ERISA statutes, and that protection under the Americans with Disabilities Act is incomplete (absent “subterfuge,” selective insurance underwriting is not prohibited discrimination).

After the collapse of the Clinton health reform efforts in 1993, and the shift to Republican control of the House of Representatives in the 1994 election, the 1995/1996 congressional term saw the introduction of new health (and genetic) privacy legislation. Four pieces of legislation introduced that session addressed issues of genetic privacy and discrimination, and the issue was addressed during committee hearings on the Health Insurance Reform Act of 1995, introduced by Senator Nancy Kassebaum (R-KS).

This last development is significant, because the Health Insurance Reform Act of 1995 was a major component of what became the Kassebaum-Kennedy Health Insurance Reform Act of 1996, ultimately adopted as the Health Insurance Portability and Accountability Act of 1996 (HIPAA). While the major purposes of HIPAA were to “reform” the small group and individual health insurance markets nationwide, this effort mostly has been unsuccessful in that policy-making sense. The most far-reaching effects of HIPAA have come from Title IV of the Act, which established the regulatory procedure and authority for the Department of Health and Human Services to adopt what became The Standards for Privacy of Individually Identifiable Health Information (“Privacy Rule”) *if* Congress did not adopt medical privacy legislation within three years of HIPAA’s enactment on August 21, 1996.

Congress did not do so, and so during the Clinton administration, HHS proposed a Privacy Rule and released it for public comment on November 1999. The Department received over 52,000 public comments and adopted a final rule published (effective) December 28, 2000. In March 2002, under the Bush administration, HHS proposed and released for comment modifications and a proposed, new “final” Privacy Rule, and received over 11,000 public comments. The new version of the Privacy Rule was published August 14, 2002, and became the Privacy Rule effective for regulatory purposes.

In its report on the Health Insurance Reform Act of 1995 (the original Kassebaum bill) the Senate Committee on Labor and Human Resources clearly noted that for policy reasons, “health status” and “medical history” covered by the act should be read broadly to include genetic information. As such, the rule-making process that led to the Privacy Rule could have – in classic subsystem operation – resolved many of the concerns regarding genetic privacy. Ultimately, this did not happen. The Privacy Rule as adopted, after an extensive comment process, excludes a group health plan with less than 50 participants administered solely by an employer, but more importantly it excludes from protected health information “employment records that a covered entity maintains in its capacity as an employer.”

While the rules could have been drafted to address broader health insurance and employment privacy and discrimination concerns, they ultimately were not. This circumstance led directly to the introduction by Representative Slaughter (D-NY) and Senator Snowe (R-ME) of the Genetic Information Non-Discrimination Acts of 2003, 2005 and 2007. Further analysis of these events demonstrates how advocacy coalitions attempted to work within the political subsystem on both sides of the issue to influence the ultimate policy outcomes in the HIPAA Privacy Rule, and how in the aftermath, genetic privacy advocates have found policy entrepreneurs to serve as their allies in an attempt to resolve these issues at the macro-policy level.

### **GINA and the Politics of the Policy-Making Process**

In April of 2007, immediately following House passage, Senator Kennedy issued a report on the Genetic Information Nondiscrimination Act of 2007 as Chairman of the Senate Committee on Health, Education, Labor and Pensions (the HELP Committee). This extensive report tracked the origins and history of this legislation from the early 1990s forward.

The report identified the purpose of GINA as:

The purpose of this legislation is to protect individuals from discrimination in health insurance and employment on the basis of genetic information. Establishing these protections will allay concerns about the potential for discrimination and encourage individuals to participate in genetic research and to take advantage of genetic testing, new technologies, and new therapies. The legislation will provide substantive protections to those individuals who may suffer from actual genetic discrimination now and in the future. These steps are essential to fulfilling the promise of the human genome project.

As to health insurance, GINA would prohibit discrimination in health insurance in employer-sponsored group plans, health insurers in the group and individual policy markets, Medigap and state and local (non-Federal) government health plans. GINA would increase existing (ERISA) protections and supplement other protections to restrict the use of genetic information obtained by insurers in processing reimbursement. GINA would further prohibit insurers from requiring genetic testing and would restrict the use of genetic information in insurance underwriting.

As to employment GINA prohibits the use of genetic information in employment decisions by employers, unions, employment agencies and training programs. If an employer obtains genetic information it shall be considered confidential and treated by the employer as such.

These protections, the Kennedy Report notes, are about perception as much as reality; there is no real documentation of discrimination in insurance or employment, rather the “expressed belief” is documented in a series of surveys that insurers and employers *will* discriminate if they gain possession of genetic information. Some legal protections currently exist, but there are gaps in federal legislation and in the availability of existing state protections.

The HELP Committee of the Senate has been conducting hearings on genetic nondiscrimination since 1995. In 1996, as part of the passage of HIPAA the Senate prohibited discrimination against any individual enrolled in a group health plan because of health status, including health status based on genetic information.

HIPAA did not directly address medical privacy; the legislation provided that HHS must issue regulations on comprehensive medical privacy if Congress did not pass comprehensive medical privacy legislation by August 21, 1999. Congress did not do so, although the Senate HELP Committee continued to hold hearings on genetic discrimination in health insurance and employment. After the adoption of the HIPAA Medical Privacy regulations by HHS, the HELP Committee and the Senate as a whole have considered genetic nondiscrimination legislation which is broader in application than the HHS regulations, as noted in earlier discussion of the Genetic Information Nondiscrimination Act of 2007.

### **Policy-Making Theory**

Several academics (Salisbury (1992); Berry (1997); Browne (1998)) have observed that the activities of interest groups, and their impact on federal policy-making, began to shift significantly in the 1980s. Correspondingly, the actions of members of Congress and of the administrative bureaucracy have shifted significantly as well. Coalitions are much more

important than prior to the 1980s. In illustrating modern policy-making, the Advocacy Coalition Framework (ACF) is a very productive tool.

Over twenty years ago, political theorist Terry Moe posed what he viewed as a central question in American political analysis: how do you construct a proper theory of political organization and structural choice in the context of political uncertainty? As he noted, the problems are many and the task is complex. Most critically, we must attempt to explain policy change and learning through examination of the reality of political organization and structural choices (Moe, 1980).

Also twenty years ago, Paul Sabatier first described the theoretical structure he labeled the Advocacy Coalition Framework (ACF). There have been other theories of the policy process proposed and developed over the same twenty year period; policy theorizing of this type has blossomed. But through the evolution of thinking about the ACF, Sabatier and others – most notably Sabatier’s partner Hank Jenkins-Smith – have developed one analytical tool to use in answering Moe’s central questions (Sabatier, 1993; Sabatier and Jenkins-Smith, 1993; Sabatier, 1999; Sabatier and Jenkins-Smith, 1999).

### **Policy Change in the ACF**

Sabatier has a central purpose of his own in developing a theory of the policy process: “The process of policymaking includes the manner in which problems get conceptualized and brought to government for solution; governmental institutions formulate alternatives and select policy solutions; and those solutions get implemented, evaluated and revised ... Given the staggering complexity of the policy process, the analyst *must* find some way of simplifying the situation in order to have any chance of understanding it.” And so, he argues, we have a need for theories of the policy process – ways of simplifying the manner in which we look at the process of policy-making in order to better understand it.

Since the original 1988 article in which Sabatier proposed the ACF, he and Jenkins-Smith have continued to develop this “theoretical lens on public policy,” revising it every “six years or so (1993, 1999, 2006).” The ACF views the central element of policy analysis to be the examination of “policy change,” the evolution of policy-making over time. This encompasses the “formulation, implementation and reformulation of policies through the process of “policy-oriented learning. “

The locus is actors in the policy subsystem, dealing with a policy issue over a period of a decade or more. These policy actors form the advocacy coalitions that are at the center of the framework. Each coalition has a central, shared belief system, a “set of basic values, causal assumptions, and problem perceptions”, and each coalition, based upon this shared belief system, shows a “degree of coordinated activity over time.”

The primary task is to “identify the conditions under which a productive analytical debate between members of *different* advocacy coalitions is likely to occur.” This examination focuses upon three variables: 1) the level of conflict over an issue 2) the “analytical tractability” of the issue and 3) the nature of the subsystem itself.

Recognition of the stability of core belief systems and the centrality of these belief systems to policy actors is also a critical element of the ACF.

### **Policy Subsystems and the ACF**

The ACF focuses analysis first on the institutional setting for action: the policy subsystem. Focusing on this setting helps explain changes in beliefs and policies among actors, as well as the manner in which actions are “shaped and constrained” by larger governing systems in which political subsystems are placed.

A useful way of looking at governing systems is in terms of macro-policy or “high politics” systems, below which are policy or micro-policy subsystems. Major decisions that may change the political power structure in a major policy area are macro-policy decisions which the ACF has labeled external system events. These decisions often result from an inability to resolve policy issues at a lower level.

Micro-policy systems are “relatively hidden” policy systems where government policies of limited public interest, often in areas of technical complexity, arise. At this level are networks of policy actors and decentralized power structures. Communications are informal, but frequent among subsystem actors, which include interest group representatives, government officials and their staff, bureau and agency personnel, non-governmental policy specialists, and interested media members.

These policy subsystems are a form of “functional representation,” and policies of regulation or redistribution that can be resolved without resort to new macro-political legislation are addressed at this level. It is at this level that interest group conflict – through the actions of advocacy coalitions – most frequently manifests itself. A subsystem can be “dominant,” that is controlled by a small number of actors with significant influence and control over policy outcomes, with stable relations among policy actors. Alternatively a subsystem can be “competitive,” with coalitions in constant competition for control.

Recent work by political scientists of the historical institutionalism school is consistent with this aspect of the methodology of the ACF. Kathleen Thelen (1992) and Margaret Weir (1992) have argued respectively that “institutions evolve (with) shifts in the political coalitions on which they rest to inspire or compel changes – sometimes abrupt and discontinuous but more often incremental and cumulative,” and that historical attention is needed to “the organizational substructure of politics, particularly to processes of issue definition and coalition building among nonelite actors.”

These observations are particularly apt given Moe’s work on political uncertainty and structural choice. The most significant issue, Moe would argue, is “not simply to get the policies and structures (an interest group) wants in the current period, but also to design them in such a way that they have the capacity to survive and prosper in an uncertain political future.”

### **The Organizational Politics of Actors, Interest Groups and Coalitions**

Actors within policy subsystems often have multiple functions, operating as actors with political expertise and beliefs, as representatives of interest groups or other identifiable organizations

operating within the subsystem and acting as advocacy coalition members. In these capacities actors seek to influence policy, to avoid political uncertainty – or at least to cope with it – and to influence structural choices. All of these functions are influenced, in ACF theory, by each actor’s belief systems, and by the organizational and interest group dynamics that are tied to the formation of advocacy coalitions.

In the American system of politics, collective opposition to change is easiest to organize and advance. Action to effectuate change is much more problematic; it requires agreement on purpose, goals and tactics. As each of these elements becomes more specifically focused, more energy and resources have to be expended in agreement on a common purpose and a course of collective action. For instance, contrast the concerted efforts in opposition to the Clinton administration’s 1993 attempts at health reform (see Chapter 5 of Theda Skocpol’s *Boomerang: Health Care Reform and the Turn Against Government* (1997)) with the several decades of effort by earlier reformers that led finally to the 1965 adoption of Medicare (see Chapters 1-5 of Theodore Marmor’s *The Politics of Medicare* (2000)).

As the ACF has evolved, there have been attempts to address these collective action problems. According to ACF theory, collective action is enhanced by sharing the expense of a common effort to effectuate change (the “transaction costs”), and a common enemy tends to create allies. Policy change is much easier to resist or deflect than to accomplish. Regulation is more easily effected than re-distribution, and “self-contained” regulation (such as legislation of prohibition) is more easily effectuated than regulation by bureaucratic activity over time. A certain amount of political uncertainty can be removed through “self-contained” regulation, as it tends to set into place what might be called “political property rights.”

### **The Input of Policy Entrepreneurs**

Policy Entrepreneurs (PEs) are policy actors that propel political and policy changes. They transform existing coalitions and add new dimensions to policy debates in the effort to effect policy change. In many instances this function will be served members of Congress who decide to take on “national” issues, issues with a scope that extends well beyond the interests of their immediate constituency. In national policy-making it is not necessary to be in Congress to serve as a policy entrepreneur, but members of Congress are uniquely positioned to take on these tasks.

These efforts are critical in defining ideas and moving them through the policy process, looking for opportunities to effect positive policy change and seizing these opportunities when presented. PEs “frame” an issue to move it to the proper level of attention in policy-making institutions together with the interest groups that they mobilize or with which they ally. They interject “new dimensions of evaluation” into otherwise conservative policy institutions through what Schattschneider (1960) first labeled “conflict expansion.”

For conflict to “expand” to the level necessary for policy change to occur, “new definitions” of policy problems and solutions must be interjected into the policy process. New interests will be activated to participate in the policy process as the conflict over the need for policy change “expands.” In cases where subsystem control of an issue serves to inhibit policy change, PEs redefine the issue to de-stabilize subsystem control. Redefinition serves the purpose of reframing issues in familiar terms and values such as “privacy” or “nondiscriminatory” or “equitable.”

The ACF focuses much of its attention at subsystem levels of policy-making in order to provide an analytical framework that can view policy change as a product of a process that occurs over as much as a decade or more. At the same time, the ACF recognizes that policy change happens for a reason, not for its own sake. Either the activities occurring within a political subsystem gradually reflect a recognition that policies need to be changed or at least adjusted (policy learning), some significant “shock” external to the subsystem literally forces change to occur, a shock “internal” to the subsystem precipitates change through the disruption of control by a dominant coalition, or the actors within a subsystem negotiate change.

One other circumstance can cause policy change to take place: the inability of subsystem actors to effect necessary changes leads policy entrepreneurs to seek change at another level of policy-making.

When we examine the case of the Genetic Information Non-Discrimination Act, we will see an example of a perceived need for change that advocacy coalitions could not resolve at the policy subsystem (in this case administrative agency) level. This being the case, the contest over policy change has moved to Congress, in part because change has been effectuated in no other way.

### **The ACF and Interest-Group Theory**

Consideration of policy-making is helped by an understanding of two additional concepts: interest-group theory and agenda-setting. Questions have been raised about the “assumption” implicit in the ACF that collective action by members of an advocacy coalition is a given. Interest – group theory will help resolve these collective action concerns, and also help provide a link to an understanding of the nature of agenda-setting.

A connection has been formed between the study of interest groups and the ACF in the work of Andrew McFarland. McFarland’s work on the political process theory of interest groups in a pluralist system has been developed over almost forty years (1969 to present), finding its most thorough exposition in his 2004 book *Neopluralism, the Evolution of Political Process Theory*. As noted in *Neopluralism*, while a great deal of work has been done in the study of interest groups, the classic focus has been on why individuals join groups and how groups maintain an organizational structure. McFarland notes that Baumgartner and Leetch, in their 1998 book *Basic Interests: the Importance of Groups in Politics and Political Science*, argued that more theoretical work should be directed to the study of the “role of interest groups in the policy process.”

For twenty years, McFarland has argued that the best way to understand the role of interest groups in the public policy process is through the application of the theory of triadic power. Under this theory – adapted from the work of James Q. Wilson (1973) – the government policy process takes place in specific policy areas. Economic producers (P) organize to lobby for rents in their area of economic production. Countervailing interests (CV) organize to compete with and oppose the interests of P. Administrative agencies with greater or lesser autonomy (AA) control much of the policy process in each area of production, and the unit formed by these three interests creates a “power triad” in the area of production.

McFarland's theoretical work has evolved from and expanded on the concept of triadic power, which is still at the core of his work; added to this are the concepts of routine politics and high politics, and the idea that the focus of the policy subsystem "cycles" from one to the other. Routine politics is the normal, day-to-day decision-making and administration within a policy area. High politics is the political process of policy or administrative structural change.

McFarland's concept of interest-group cycles is adapted from Schlesinger's thinking in *The Cycles of American History* (1986). McFarland's basic argument is that the actions of interest groups, in addressing specific issue areas, go through cyclical phases. In periods of routine politics producer groups are commonly in control, leading to periods of excessive control and leading to popular discontent. This is followed by a transition period during which countervailing power groups coalesce, followed by a period of high politics in a reform cycle. During reform cycles countervailing power groups and autonomous government agencies respond with corrective policy-making, after which another transition phase occurs in which matters recede from the public agenda, producers maintain their constant efforts for control and another era of producer dominance follows before the cycle revolves again.

In *Neopluralism*, McFarland brings his study of interest group participation in the policy process to fruition. In doing so, he finds the ACF to be the most useful theory for purposes of studying how interest groups participate in, and influence, the policy process. Political process theory, McFarland argues, should address three questions. Who has the power here? How is policy made in this area? What are the activities of interest groups in this area? The answers, he argues, are best arrived at by recognizing the power of an analysis using the triadic concepts, agenda-setting as seen through the interest-group cycle theory, and the tools of the ACF.

In applying the ACF to interest groups, McFarland first observes that the ACF acknowledges the utility of Hugh Hecho's (1978) concept of issue networks which McFarland refocuses as "policy networks." A policy network is an ad hoc communication network of interest group participants, bureaucrats, public officials, academics and interested media members interested in a common set of policy areas. Secondly, from within a policy network, and remaining relatively stable over a long term, are advocacy coalitions. These operate to influence the processes within policy subsystems. The common beliefs that hold advocacy coalitions together are not only material interests as Sabatier and Jenkins-Smith had noted (and McFarland reinforces), but extend to core social and political values as well.

### **Agenda-Setting, Interest Groups and the ACF**

Interest-group cycles theory is a theory of political agendas. One of the driving forces in agenda-setting as it affects interest groups is the impact of political cycles on the content and timing of policy agendas. John Kingdon's influential theories of agenda-setting utilize a policy network concept as well, labeling these networks "policy communities." In Kingdon's work (1995), the existence and activities of these policy communities becomes critical to problem recognition, problem definition, the generation of policy proposals, and the selection of policy alternatives. The ultimate question becomes, is there a problem that needs a solution?

Conceptually, McFarland notes, this is consistent with ACF theory. The evolution of the perspectives of members of a policy network or community is, Kingdom recognizes, one of three

major contributors to the setting of governmental policy-making agendas, along with significant, or triggering” public events that accelerate problem recognition, and the effects of political events and processes.

Some policy communities, Kingdon observes, are close knit and some are more fragmented. A close-knit policy community shares a commonality of outlook, orientation, vision; a fragmented community tends to be relatively unstable and lack a shared sense of structure. Close-knit policy communities have a greater impact on the policy agenda, and fragmented communities less so. But surrounding each “area of policy concern” (Kingdon’s description) – the policy subsystem, where policy problems emerge and are matched with alternative policy solutions – such a community exists. And in turn, as problems emerge and rise higher on the public agenda, the impact of the interested policy community on resulting policy outcomes becomes more significant.

Kingdon poses a straightforward question: How does the list of potential alternatives for public policy choices get narrowed to the ones that actually receive serious consideration? There are, he says, two classes of answers: alternatives come to life and evolve in what he calls the “policy stream” (again, analogous to the policy subsystem), and it is policy specialists that work within a particular “policy area” (again, a subsystem) that are engaged in the narrowing process.

Kingdon describes three agenda-setting and alternative-selection processes through which policy-making will occur: “the inexorable march of problems pressing on the system,” “the gradual accumulation of knowledge and perspectives among the specialists in a given policy area,” and “political processes.” The second of these processes would originate in a policy subsystem. Policy-making through the other two processes would originate at the levels earlier described as macro-political. At all three levels McFarland’s cycles of excess and reform, routine and high politics, will occur.

When Congress is involved in the policy-making process some additional agenda-setting elements will apply as well, including some elemental agenda differences in the Senate and the House. These will become relevant to the analysis in due course.

### **Agenda-Setting in the U.S. Senate**

One of the early, pre-eminent observers of interest groups was Jack Walker (See “The Origin and Maintenance of Interest Groups in America,” *American Political Science Review* 77: 390-406 (1983). In his 1977 article, “Setting the Agenda in the U.S. Senate: A Theory of Problem Selection” he addressed the significant restrictions on the opportunities for members of the US Senate to move individual agenda interests. These opportunities are restricted by a variety of factors:

The Senate's capacity to shape its own agenda is increasing, but members are still able to exercise little discretion over the scheduling of items for debate. Much of the business transacted by the Senate is either mandated by the Constitution or required for the maintenance of the vast federal establishment. Each year a budget must be assembled, innumerable amendments made to existing statutes, and presidential appointees confirmed or rejected. In addition, the daily schedules of

individual Senators are jammed with activities – subcommittee hearings, talks with constituents, lobbyists or reporters, roll calls on the Senate floor, consultations with staff members – that originate with other people and are virtually unavoidable. Little time and energy remain for reflection or the promotion of new legislative departures (Walker, 2007).

In addition, Walker points to what he labels “sporadically recurring problems” – issues that must be addressed because of prior legislation (typically reauthorization items) or administrative oversight. The Senate, Walker observes, tends to respond even more than the House to the exigencies of the moment, leaving little opportunity to address the type of priority that GINA represents: establishment of broad policy statement, particularly in the absence of a perceived crisis. Walker has posited three “features or conditions,” the presence of which would tend to significantly increase the probability of a legislative item successfully appearing on the Senate’s discretionary agenda: actual (not theoretical) impact on large numbers of people, convincing evidence that a serious problem will be addressed (again, actual not theoretical) and an easily understood solution exists, has been identified and is being adopted.

If all three of these desirable characteristics are clearly present, the likelihood increases significantly that the matter will get space on the Senate’s agenda, though this is by no means guaranteed. Matters not meeting all of the observed elements have virtually no chance. Senator Kennedy’s report, discussed earlier, virtually establishes the failure of GINA to meet Walker’s observed agenda-setting standards: the problem that GINA was designed to address was prospective and theoretical, not representative of a clear and present necessity.

### **Looking at GINA with a Theoretical Lens**

The Council for Responsible Genetics is a genetic professionals group formed 25 years ago to “foster public debate about the social, ethical and environmental implications of genetic technologies.” As such, it is an advocacy coalition with a set of beliefs generally consistent with efforts to “work through the media and concerned citizens to distribute accurate information and represent the public interest on emerging issues in biotechnology.”

A major player within the Council for Responsible Genetics, and in seeking passage of GINA, was the Genetic Alliance. In describing itself, the Genetic Alliance says that it is “a coalition of more than 600 advocacy organizations serving 25 million people affected by 1000 conditions.” The existence of the Genetic Alliance is premised on the legitimacy of the coalition perspectives discussed thus far. Founded 20 years ago, the Genetic Alliance was intended to facilitate the formal linkage of what has grown to over 600 organizations, each of which constitutes an interest group dedicated to building the capacity of these organizations to address issues of concern to “the genetics community.”

Many of the advocacy organizations that had joined the Genetic Alliance had been formed to link together people with a personal interest in defined genetic diseases that - while devastating in their effects on sufferers – impact very limited portions of the US population. By themselves these groups had limited resources and found it virtually impossible to impact broad public policies. But 20 years ago the organizers of the Genetic Alliance could see the potential for

significant developments in genetic research, and the attendant emergence of a wide array of policy issues that would be of significance to each of these smaller advocacy groups.

The Genetic Alliance openly avows “leveraging the voices of its members in formal communication with governmental officials and agencies” and the “application of organizational network theory” in advancing the interests of its member advocacy organizations. The Genetic Alliance is literally an advocacy coalition as defined by Sabatier, organized around a central, shared belief system. The Alliance has enumerated this shared belief system, in its mission statement, its statement of its central philosophy and consistently in its position statements over the course of its history.

Both of these coalitions joined in an open fight against what they labeled “genetic discrimination.” What is “genetic discrimination” as these coalitions have defined it in this policy dialogue? An employer’s use of genetic information to make hiring and firing decisions would be genetic discrimination. An insurance company’s denial of coverage based on genetic test results would be genetic discrimination. These coalitions centered themselves around a belief that genetic information must not be used to discriminate in insurance underwriting or in employment decisions.

GINA was the subject of discussion and public dialogue among many of the small groups and the researchers which were first concerned about genetic discrimination, and the establishment of genetic privacy, in the late 1980s and early 1990s. In classic fashion issue networks emerged surrounding the genetic research that was being done at the time, as it began to show promise and receive public funding. Through the development of policies by the Equal Employment Opportunity Commission (EEOC) under the Americans with Disabilities Act, the enactment of HIPAA, the medical privacy Final Rules under HIPAA and the debates over GINA 2003, 2005, and 2007, advocacy coalitions have operated within classic issue networks centered on genetic issues.

### **The Continuing Call to Arms**

The ACF is a model that supports, and conforms to, much of the best work on interest group theory, in that its primary focus is on the coalition-building that is most influential at the administrative level. Moreover, as a model of the policy process, the ACF emphasizes the difficulty of influencing policy-making at the Congressional level by any means, including coalition-building. As long observed by political observers, coalition-building can best marshal forces at the Congressional level to create opposition to the adoption of new policies.

The interest groups that had set passage of GINA as a priority had to continue to try and influence Congress in a positive way. But as interest group theorists and application of the ACF demonstrate, the task at hand was a daunting one. Robert Salisbury and Jeffrey Berry, in contemporaneous articles in 1989 and 1990 both observed that the sub-governments and iron triangles that political scientists had observed from after World War II until the mid-1970s had begun to disappear in the early 1980s, replaced by the issue networks discussed earlier. And both observed, as Salisbury put it, that the result was a substantial increase in interest group activity in Washington with less, not more, impact on Congressional policy-making. And as William P. Browne documented in the early 1990s, a further result has been that members of Congress have

reverted, if you will, to being most concerned about constituent issues and not broad policy issues that don't sell "back home."

These were the impediments that the proponents of GINA faced. Attempts to gain passage of the type of broad, general policy-making that GINA represented are daunting, even if the nature of the policies in question would seem to be lacking in controversy.

### **How GINA Came to Pass into Law – and What We Can Learn**

The proponents of legislation that would create a national policy of genetic privacy and prohibit genetic discrimination mounted what turned out to be an almost two-decade effort to accomplish this end. First, those that had similar interests in the passage of such legislation began public dialogue about the issue in classic issue networks. There was opposition over the years from business and insurance interests, though not in a consistently organized fashion due to the lack of any apparent agenda-setting event that would catalyze an effort to make genetic discrimination issues emergent.

Still, citizens groups concerned about a myriad of possible genetic-based medical advances aligned with researchers concerned with the need for research volunteers who were not inhibited by issues of genetic discrimination to argue for protective legislation. The arguments were first centered at the political subsystem level of administrative agencies (EEOC, HHS), and then moved to the level of Congressional macro-politics as the proponents gained more and more converts, eventually reaching to members of Congress.

This is the policy change that Sabatier and McFarland have theorized about in arguing the merits of the ACF as a method for analyzing the evolution of public policy. An examination of the long development of the policy momentum that led to the passage of GINA in the 110<sup>th</sup> Congress would seem to bear out the legitimacy of the ACF.

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## Genetic Services Policy Project Final Report

### **Appendix C: Case Study of Intermountain Healthcare’s Clinical Genetics Institute**

#### ***Why the Case Study Approach?***

The field of genetic services is a relatively new one; some organizations are embracing rapid growth in this area while others are moving more slowly. In such a new and varied field where there is as yet no standard response, it is helpful to use a case study approach to examine clinical integration of genetic services. The primary advantage of this approach is that the smaller unit of analysis allows for an exploration that is more detailed, more specific, and therefore more meaningful. It is also useful in facilitating examination and analysis at the level of the organization. Our purpose is to study the integration of genetic services into clinical settings.

In selecting a site for a case study, we sought organizations that are intentionally integrating genetic services into their clinical pathways. Intermountain Healthcare, located in Salt Lake City, Utah, provides many useful characteristics for a case study. The city itself, and Utah generally, is particularly conducive for such research because of several factors: the strong cultural interest in family history, the availability of an extensive body of medical records linked to family pedigrees, public willingness to participate in biomedical and public health research, investment in biotechnology as an economic base, and the presence of the University of Utah and various commercial enterprises. “Because Utahns tend to have large families and keep extensive genealogy records, they are ideal partners for investigating human genetics” (Eccles Institute of Human Genetics website). In addition, the traditionally low rates of tobacco and alcohol use and relatively homogenous demographics of the population minimize variation of many of environmental and socioeconomic contributors to disease and provide “a much cleaner data set,” according to University of Utah instructor Deborah Neklason. “It means [researchers] can more easily separate genetic influences from other, lifestyle-related causes of illness” (Sussingham).

Intermountain Healthcare was selected as the site of the case study largely because it is a leading integrated health care system that is interested in genetics. Intermountain began exploring means of integrating genetic services in 2004, culminating in the creation of the Clinical Genetics Institute (CGI). We chose to use CGI as the basis for a case study in part because of its surrounding conditions and a commitment by top administrators to integrate genetic services in an explicit program. In addition, Intermountain’s structure as a payer and provider allows for the study of multiple facets: as the payer and provider, as the payer for other providers, and as the provider for other payers. The fact that Intermountain offers both clinical care and insurance products provides an opportunity to study the integration of genetic services from the perspectives of health care providers, hospital administrators, and payers.

#### ***Narrative about Utah and Intermountain***

The state of Utah is an interesting outlier in statistical measures, a state in which the population tends to be younger, more educated, and healthier in many regards than the national averages. The population has a higher percentage of Caucasians—in fact, Utahns of northern and western

European ancestry were the population chosen to represent Caucasians in the International HapMap Project (National Human Genome Research Institute, 2006)—a strong cultural interest in genealogy and willingness to participate in research, and widespread economic investment in the life sciences and biotechnology industries. Salt Lake City is home to the state government, a public university, many for-profit businesses and nonprofit organizations, the Church of Jesus Christ of Latter-Day Saints (often referred to as the LDS or Mormon Church), and Intermountain Healthcare. The teachings of the LDS Church—the importance of genealogy and family history, healthy living, education, and volunteering—are reflected in many of these anomalous trends.

While traditionally home to a relatively homogenous citizenry of Caucasian residents, conservative values, and lower than average cost of living and wages, things are starting to change. Immigration, cost-of-living, and salaries are increasing (Key Informant A, 2006), and liberal views are becoming more common in the urban area (Key Informant B, 2006). Salt Lake City, the capital, seems to be changing more quickly than the rest of the state. Divides in political views, coupled with the competing priorities of life sciences, infrastructure development, and education, may lead to tensions in the future over the proper role of genetic services.

The state government is conscious of Utah's relatively high birthrate and how that affects both the recently expanded newborn screening panel and Medicaid budgets. Medicaid finances nearly one-third of the state's births, which is lower than the national average of more than 40 percent (Statehealthfacts.org website). Disproportionate shares of Medicaid enrollees in Utah live in a few outlying communities comprised mainly of members of the Fundamentalist LDS Church. FLDS members often practice plural marriages (polygamy) and family intermarriage. The twin towns of Hildale, Utah, and Colorado City, Arizona, have higher rates of poverty than their state averages: 33 percent of city residents receive food stamps, compared to less than 5 percent in Utah and 7 percent in Arizona (Zoellner, 1998). The cities also have the world's highest incidence of fumarase deficiency, a rare genetic disorder that causes severe mental retardation, which genetics experts attribute to the practice of cousin marriage (Szep, 2007).

There is growing interest in research into birth defects and the genomic contribution to chronic disease; the University of Utah is conducting bench science in this area. In 2006, the U.S. Centers for Disease Control and Prevention awarded a \$2 million, five-year grant to a new Intermountain Center of Excellence for Infection Prevention Strategies (INTERCEPT), a partnership between Intermountain Healthcare, University Health Care and the Veterans Affairs Salt Lake City Health Care System, for a joint study on health information technology and infection prevention (University of Utah, 2006).

Intermountain Healthcare, with its companion insurer SelectHealth, is one of the largest health systems in Utah, and one of the top-rated integrated health care systems in the country. Despite occasional concerns about market share and antitrust regulations, the state generally perceives Intermountain in a positive light. Given a growing national interest in genetics in the last decade, Utah's rich and unique genetics-related resources, and an integrated data system, Intermountain Healthcare saw an opportunity to develop genetic services capacities with an emphasis on prevention, clinical utility, and cost-effectiveness.

### ***The Clinical Genetics Institute***

Intermountain Healthcare has been working to integrate genetic services into the broader health care delivery system through its Clinical Genetics Institute (CGI). In spring of 2004, several Intermountain executives began discussing the idea of such an institute. The Clinical Genetics Institute was the brainchild of three Intermountain Healthcare physicians and was supported by the administration. Some touted genetics as the “next big thing,” while others described it as “the ultimate preventive medicine” (Key Informant C, 2006). Despite an enthusiastic task force at the beginning of the process, within a year one of the three promoters passed away and one retired from active practice. Although the institutional support for CGI remained, the clear vision and focused energy were lost. The administration hired a director with prior experience in genetics clinics, who brought his own vision of how best to integrate genetic services.

CGI’s past: During key informant interviews in the summer of 2006, we discovered that there is little consensus on CGI’s original purpose and that its role within Intermountain was not clearly articulated in the beginning. One member of the senior leadership team recalled that the purpose of CGI was to do genetic research and discovery to strengthen clinical care programs, and to serve as the broader base of genetics information. Another leader believed CGI was created both to serve as a central genetics resource for Intermountain and to evaluate genetic tests.

CGI in the present: We first interviewed key informants during the summer of 2005. Little was known about the Clinical Genetics Institute, which had only begun a few months earlier. During our expanded follow-up interviews in the summer of 2006, we noted no new large-scale activities. There was, however, the addition of the Adult Genetics Clinic, a monthly clinical diagnosis activity within CGI. We found that, within and outside of Intermountain Healthcare, knowledge of CGI’s activities and purpose was limited and often conflicting.

In an attempt to evaluate whether the CGI had a significant impact on clinical services, claims data were also reviewed. Specifically, data were compiled based on CPT codes (service codes) for genetic testing, genetic counseling and consultation for conditions consistent with the clinical case studies used in this project (e.g., breast/ovarian cancer, cystic fibrosis, sickle cell disease and multiple congenital anomalies) over a three year period (calendar years 2004-2007). Analysis of these data revealed no significant changes in service utilization or claims over this time frame. It may be that more time would be necessary to see any service trends and/or these data accurately reflect the changing focus of the expectation of the CGI.

Although most people within Intermountain Healthcare spoke highly of CGI’s leadership, few were able to articulate its specific activities, and many outside stakeholders had not heard of CGI. One Intermountain senior manager thought CGI’s primary purpose was to evaluate the counseling and payment issues surrounding new genetic tests and to develop protocols for clinical use. The respondent, who did not know any details, thought of CGI as a “system interface” for genetic activities and speculated that it might be working with the oncology program. The respondent believed that CGI’s impact lies in helping to surface the future of genetics across the Intermountain system, facilitating genetic counseling in the oncology program, helping people understand the need for guidance regarding new genetic tests, and serving as the impetus for enacting that structure. The staff of CGI saw its role as promoters of genetic services, though the original plan for CGI was to provide critical technology assessment.

When asked whether CGI is meeting expectations, a senior manager responded with uncertainty as to what to expect, noting that things are moving “at a snail’s pace” (Key Informant C, 2006). However, the manager also recognized that it can be difficult to get people involved and was pleased with the progress CGI has made so far. Another manager, responding to the same question, answered that CGI seems to be consistent with company expectations but was not personally sure what to expect. Representatives of the affiliated insurer, SelectHealth, seemed to have a more specific idea of CGI’s activities, a view that was not echoed by other groups and individuals. They saw a distinction between the types of genetic test analyses performed by CGI and SelectHealth. There is a difference between the payer perspective of cost-effectiveness and the clinical perspective of clinical utility; SelectHealth worked with CGI to align benefit design with clinical needs. SelectHealth representatives described their relationship as “cordial and collaborative” (Key Informant D, 2006). Others with whom we spoke did not seem to know what CGI does. Health plan physicians saw CGI’s role as educating physicians once technology assessment decisions were made.

CGI’s future: One senior leader envisioned CGI playing a larger clinical and basic research role, performing crosscutting education, and conducting technology assessments in the future. Another manager predicted that CGI’s role would be as a collaborator, potentially with the University of Utah or Sorensen Molecular Genealogy Foundation, and would offer education “in a big way” (Key Informant C, 2006).

The three original promoters of CGI envisioned that it would conduct research to guide the policy of Intermountain and SelectHealth in using new genetic tests. The director, however, developed a broader set of activities for CGI that was more in line with his interests and experience; the shift in focus coincided with the center’s change in leadership. Without the involvement of two of CGI’s original promoters, the driving impetus became less immediate and the focus less clear, leaving CGI staff to figure out their place within the broader organization. As of the 2006 interviews, CGI had not reported to the Board formally and had only talked informally with an immediate supervisor on a somewhat regular basis. The supervisor had a generally positive view of CGI, but did not keep track its actual activities. As in most organizations, this case illustrates a few basic principles: communication is key; commitment of leadership is necessary; and the mission and vision must be clear and consistent.

CGI faces an organizational/structural difficulty because of the fact that most clinical programs at Intermountain are organized vertically, yet CGI is conceptually horizontal, cutting across many programs. Its current strategy is largely to be present, to attend meetings and present ideas. In particular, CGI has recognized the inadequate supply of genetic counseling and has been a champion of additional genetic counselor capacity.

The intent of the case study was to analyze how one organization worked to integrate genetic services into the health care delivery system. By examining both the Clinical Genetics Institute’s challenges and opportunities, other organizations will be better able to anticipate potential obstacles, capitalize on opportunities, and differentiate which factors are specific to CGI and which are more universally applicable.

### ***Challenges for the Clinical Genetics Institute***

If the Clinical Genetics Institute may be viewed as a fledgling organization (though the Intermountain system itself has been in existence for decades), its staff has faced challenges similar to those of many new initiatives. Such issues include the commitment of leadership, the clarity and consistency of the mission and purpose, organizational structure and funding, communication with key stakeholders, and external factors such as the economy, legislation, and community demographics.

Internal Challenges: Among the primary challenges in a new initiative are to retain leadership support and involvement; to balance limited resources with other initiatives within the organization; to possess the full range of necessary expertise; and to negotiate disagreements with internal and external stakeholders. CGI had a strong start because of the enthusiasm of its proponents and an executive-level decision to support the start-up of CGI. Once the original backers of CGI left Intermountain, however, much of the drive dissipated. The administration did not curtail CGI, but it did not actively support it either, causing (allowing) CGI's staff to fly under the radar and work to create a niche for themselves.

The diminished institutional support for CGI did not necessarily reflect an actual reduction in interest; rather, competing initiatives garnered more attention and fiscal resources. For example, Intermountain invested in a new data system and began construction of a new flagship hospital, both of which diverted significant available funding and administrative resources. The creation of Intermountain's new data system limits the addition of family history information in the current medical record system; this has hindered CGI's plans in delivering genetic services. Additionally, CGI's resources are limited by having a small staff. Though staff members display evident enthusiasm and have relevant experience, their expertise covers only a part of the broad scope of operating a clinical genetics institute within an integrated health care system. If, in accordance with the original vision, CGI were to develop new models of education and outcomes research, it is questionable whether they would have sufficient resources and background to fully develop the research and implement its findings.

Some disagreement exists, both external and internal to Intermountain, regarding the purpose of CGI. Some perceive an overlap between CGI's mission and activities that other groups are already performing. There is also disagreement about whether Intermountain needs increased genetic counseling capacity, and where the additional genetic counselors would be located administratively and physically. The delicate balance of collaboration and competition between Intermountain Healthcare and the University of Utah's medical facilities further complicates this disagreement. The lack of clear purpose and uniform stakeholder understanding hampers efforts.

The Clinical Genetics Institute does not currently have an established, measurable set of outcomes to justify expenditures and make a case for expanded capacity. The units of assessment—number of clinical services, percent increase in patient volume, reports of technology assessments, etc.—are yet to be determined, but will soon be needed to be able to assess its impact and strategically plan for the future.

External Challenges: Other challenges arise outside of the organization's direct control, such as legislative actions, regulatory changes, and the culture of the broader community. CGI and

Intermountain Healthcare have not been immune to these challenges. Intermountain, as the largest provider of health care services and health insurance in the state of Utah, has faced periodic charges of antitrust violations. Although a 2006 legislative task force reported that Intermountain did not use unfair business practices, sensitivity about its market share remains (Argue et al., 2006). Partially in response to those concerns, Intermountain's health care and insurance plans changed their names in 2005: Intermountain Health Care and IHC Health Plans became Intermountain Healthcare and SelectHealth. One of Intermountain's challenges will be to remain cognizant of the external pressures that precipitated the renaming.

Like all health care organizations, Intermountain must operate in a changing regulatory environment. Utah is one of only a few states in the country to require genetic counselor licensure, which creates new opportunities and issues for reimbursement for licensed counselors. The original version of the law required genetic counselors to have graduated from a currently accredited program. This proved problematic, however, for those who attended a program that was accredited at the time of their graduation but has since lost accreditation or ceased operations. The law was later revised to allow for a temporary license provision.

The state legislature voted down a so-called "any willing provider" bill in 2005. Approximately half of all states have "any willing provider" laws. The law typically means that health insurers must accept contracts with any willing health care provider in the geographical service area, as long as the provider is "qualified under state law" and "willing to meet the terms and conditions set forth by the insurer" (National Conference of State Legislatures).

Intermountain's insurance side, SelectHealth, offers three network HMO plans and a fourth preferred provider organization (PPO) plan. The enactment of an "any willing provider" law would have altered the structure and dynamics of SelectHealth's plans by mandating that it open its provider networks to any willing provider.

Finally, CGI is conscious of the opportunities and limitations posed by operating within the bounds of what is culturally acceptable in Utah. Staff personnel remain mindful of the connection between genetic services and reproductive decision-making, as well as the predominantly pro-life culture in the state.

### ***Opportunities for the Clinical Genetics Institute***

Internal: The Clinical Genetics Institute has several opportunities, particularly the continued support of an administration that sees CGI as a long-term investment. CGI recently reconstituted its advisory board, creating an opportunity to clarify its mission and define its message. In addition, although the new flagship hospital, heart and lung center, and trauma and critical care facilities compete with CGI for Intermountain's internal resources, they also draw external attention to Intermountain and provide more opportunities for clinical research and integration. There are also opportunities for collaboration with the genetic counselor program at the University of Utah and the state Genetics Advisory Committee.

Another advantage is Intermountain's structure. With clinics across the state and the rise of telemedicine, CGI is well positioned to deliver clinical genetic services and counseling over a broad geographic area. Because Intermountain is an integrated system, there is a seamless

transition from primary care to specialty follow-up care. This may facilitate better integration of genetic services. Intermountain also has mechanisms for integration and provider education that surpass many other organizations. With this system already in place, CGI can widely disseminate its content more easily than if it had to create new educational opportunities.

CGI could serve as an intermediary player, encouraging Intermountain and UU to move from competition to collaboration. One interviewee suggested that CGI could provide guidance regarding insurance coverage decisions for genetic services; currently, if the UU sees an Intermountain patient, SelectHealth generally will not pay for it.

External: Utah state government activities may provide opportunities for the Clinical Genetics Institute. The reconstitution of CGI's governing body coincided with that of the state Genetics Advisory Committee. The activities of the two may provide fodder for discussions and recommendations. Additionally, the requirement for genetic counselor licensure provides opportunities to advance the field of genetic counseling, which is a key component of CGI's work. Licensure allows for the possibility that genetic counselors could bill directly for their services, which may increase reimbursement and attract more genetic counselors to the state. An influx of genetic counselors would increase opportunities for collaborative training and delivery, particularly if genetics becomes an important component in all medical specialty training programs. The resulting billing information may provide data for research into how genetic counseling is used, whether it is cost-effective, and what other effects it may have.

CGI may be in a unique position to create a business case for providing genetic services in general and genetic counseling in particular. Because of the research and clinical practice possibilities, CGI and Intermountain could provide national models for effective genetic services integration.

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## Genetic Services Policy Project Final Report

### Appendix D: The Emerging Landscape of Genetic Services

#### **Introduction**

Historically, medical genetics services focused on “genetic” diseases – diseases for which genetics is the major causal factor. Increasingly, however, genetic information is becoming relevant for common complex diseases, and provides therapeutic opportunities unrelated to inherited risk. This shift has implications for the work of genetics professionals and the knowledge needs of other health care professionals. Because many new genetic tests and technologies are already available for clinical use and many more are expected to become available in the near future, this shift also poses important challenges for health care policy-makers.

Genetic services are offered in several different locations within the health care system. For the most part, however, they are currently a component of specialty referral services. A significant portion of genetics services occur in designated medical genetics clinics, often located in academic medical centers. Major factors determining where services are provided are the clinical manifestations of the disease for which services are provided, the nature of the available treatments, current screening recommendations, and reimbursement policies.

Many early innovations resulting from genomic research will enhance currently existing genetic services, creating pressure on genetics professionals to expand their availability or delegate certain services to other health professionals. Increasingly, genomic research will also provide tests and technologies that are most appropriately integrated into primary care and other parts of the health care system. Some will challenge the current delivery of health care in fundamental ways.

#### **Current genetic services**

##### Services prompted by clinical presentation or the nature of treatment

Each medical specialty provides diagnosis and treatment of a subset of rare genetic diseases, based on their clinical presentation. Retinal dysplasias are seen by ophthalmologists, hereditary ataxias are seen by neurologists, porphyrias are seen by gastroenterologists and dermatologists, and so on. Because these disorders are rare, even within specialty care, specialists with the appropriate expertise are often located in academic medical centers or other sites of tertiary referral care. Many of these centers also have specialized units that provide care for genetic disorders that do not fall readily into a single specialty domain, such as metabolic disorders and congenital malformations. Some genetic diseases are both common enough and unique enough in their care requirements to have dedicated clinical units; examples include specialized clinics for patients with cystic fibrosis, sickle cell disease, hemophilia, and the muscular dystrophies.

The pathway to care is influenced by the type of symptoms and by treatment options. Some disorders are highly treatable – for example, multiple endocrine neoplasia type 2 (MEN 2) and hemophilia. For others, treatment has steadily improved but continues to fall short of definitive control – for example, cystic fibrosis. Treatment for many genetic disorders is still predominantly palliative – for example, the muscular dystrophies. Referral for discrete components of care may occur, such as surgery for MEN 2. Care may sometimes be provided through a partnership between a specialty center, genetics professionals and the primary care provider. In this model, the patient may be seen periodically – e.g., once a year – in the referral center for consultation, with day to day care provided by the primary care provider.

#### Services prompted by screening

- (1) *Newborn screening* Some genetic disorders require the initiation of treatment in infancy, with good prospects of a healthy outcome if treatment is started early enough. This reality led to the creation of state newborn screening programs, to identify affected infants prior to the onset of symptoms and refer them to the appropriate specialty care.
- (2) *Prenatal screening* Several genetic tests are currently recommended for routine prenatal screening, including maternal serum measures to identify increased risk for neural tube defects and Down syndrome; carrier screening for cystic fibrosis, sickle cell disease and thalassemias; and carrier screening panels for women of Ashkenazi Jewish ancestry. These tests, when positive, may be followed by prenatal diagnostic testing to determine whether the fetus is affected.

#### Genetic counseling

Genetic counseling is an important adjunct service in all of these clinical settings. After a diagnosis is made, whether by clinical presentation or as a result of screening, genetic counseling provides families with the opportunity to learn about the inheritance pattern of the disorder, the risk for other family members, and genetic testing options. Reproductive genetics is an important focus of genetic counseling services. For genetic disorders that are severely disabling and lack definitive treatment, some parents are interested in the use of genetic testing to prevent births of affected children. Clinical geneticists and genetic counselors assist parents to understand their reproductive options. Carrier tests for many autosomal or X-linked recessive diseases can identify couples who are at risk to have an affected child. Prenatal diagnosis is available for many of these genetic diseases, to determine whether a fetus is affected. These tests can be used to inform the use of selective abortion or assisted reproductive technologies, including pre-implantation genetic diagnosis to prevent the birth of a child with specific genetic disease. Care is provided through collaboration between clinical genetics and perinatal medicine; often genetic counselors are located in a prenatal clinic to assist in the delivery of these services.

Genetic counseling can also assist in identifying affected family members so that appropriate health care can be provided. For example, if a young woman with breast cancer is determined to have a BRCA mutation, other family members can be tested to determine whether they have inherited the mutation, so that appropriate cancer screening or prophylactic surgery can be offered. Similarly, a diagnosis of hemochromatosis should prompt evaluation of other family

members at risk – in particular, siblings of the affected person – so that appropriate phlebotomy treatment can be initiated.

### **Evolving role of the genetics professional**

The American College of Medical Genetics defines medical genetics as “a branch of biomedical science that studies the relationship between genes and health,” clinical genetics as “a primary medical specialty focused on health and illness of individuals and their families,” and a clinical geneticist as “a physician who specializes in genetic disorders and conditions”. Genetics professionals include clinical geneticists (MDs with clinical genetic training), nurse geneticists (RN), genetic counselors, who may have MS, or PhD degrees, and laboratory geneticists.

The role of the clinical geneticist has evolved over time and encompasses a range of clinical roles (Table, page 9). The typical example of a clinical geneticist is that of a dysmorphologist evaluating children with developmental delay and/or birth defects for genetic disorders. In fact, the clinical geneticist can also be involved in prenatal diagnosis, diagnosis and treatment of metabolic diseases, diagnosis of single gene disorders, evaluation of both children and adults with neurologic and neuromuscular diseases, cancer, or infertility, and evaluation of genetic traits involved in common diseases (Williams, 2001).

In the past, the focus of clinical genetics was to help establish a diagnosis for patients with rare genetic disorders and conditions. Even in the absence of treatment, a diagnosis can provide some benefits to the patient, including the elimination of unnecessary tests, risk information for reproduction and family members, and prognostic information (Williams, 2001). Except for metabolic diseases, treatment was not part of the role of the clinical geneticist. In recent years, clinical geneticists have become increasingly involved in the management of the patients they diagnose. There is also reasonable hope that the research focused on single gene diseases will bear fruit in more definitive treatments for at least some diseases. The recent publication of guidelines and monographs about the management of genetic syndromes speaks to this growing role (Cassidy and Allanson, 2004; Trotter, 2005; Kishnani, 2006). Clinical geneticists now help coordinate long-term follow-up, periodic evaluation for known complications, and supportive treatments.

Not surprisingly, the earliest clinical benefits of the Human Genome Project are in the form of more and better tests for genetic diseases. Diagnosis is becoming increasingly more accurate with the aid of DNA-based testing. The increasing availability of diagnostic tests for genetic disorders makes it increasingly impractical for the clinical geneticist to be involved in the provision of all such tests. Some of these tests have already been taken up by non-genetic specialists, like BRCA1/2 testing by oncologists or HNPCC testing by gastroenterologists. To ensure that the use of genetic tests by non-genetic specialists is successful, these providers need to be knowledgeable about the most common genetic conditions they face, about the interpretation of the genetic tests they use, and about the type of information and counseling needed when providing such tests (Greendale, 2001). Some specialty clinics have hired genetic counselors to provide counseling to patients undergoing genetic tests, although genetic counselors are not trained to diagnose conditions.

## Emerging genetic tests

Clinical geneticists have not typically been involved in what is now called “genomic medicine,” i.e., the use of risk information about genetic susceptibility to disease or pharmacogenomic profiles in practice. Although these tests provide information with potential implications not only for the patient being tested but also for his/her family members, such tests are usually not accompanied by genetic counseling. Practice standards, including the evidence needed to justify test use, are not yet established, nor is the role of genetics professionals, either in setting practice standards or in assisting other clinicians by means of educational efforts, consultation, or counseling.

### Pharmacogenomics

Pharmacogenomics represents one of the most promising clinical applications of genomic research. Testing for gene variants associated with drug response has the potential to improve both the safety and the efficacy of drug treatment. In the most widely anticipated use of pharmacogenomics, testing would occur before prescribing commonly used drugs to assure that the appropriate drug is chosen based on the patient’s likelihood of adverse reactions or response. The level of evidence required to justify routine use of pharmacogenomic tests is not yet established; in particular, there is controversy concerning the need for randomized controlled trials to assess the outcomes of pharmacogenomically assisted prescribing. Once this approach is determined to be useful, clinicians will need to be educated in its use.

An important question in the evaluation of pharmacogenomic testing is whether it poses significant personal or social risks. Testing is focused on informing drug prescribing, but many pharmacogenomic tests provide ancillary information, defined as information unrelated to drug response, such as predisposition to diseases for which the individual is not currently seeking treatment or does not manifest symptoms, or prognostic information that is not informative for treatment. The implications of this information for informed consent or appropriate use of pharmacogenomic tests is not yet resolved. Conceivably, some pharmacogenomic tests could pose sufficient risks to make genetic counseling a consideration.

Some pharmacogenomic tests will be introduced as a component of a new therapeutic. An example is testing *Her-2-neu* amplification in breast tumor tissue, in order to determine if the patient is a candidate for Herceptin therapy. To the extent that testing measures acquired as opposed to inherited genetic change, as in this example, the potential risks of the testing process are reduced (Haga and Burke, 2008).

### Gene expression profiling

Gene expression profiling represents another new genetic testing strategy. As with testing for *Her-2-neu* amplification, this testing approach has been used to measure acquired genetic change. For example, Oncotype Dx and MammaPrint are two currently available tests that measure gene expression in breast tumor tissue, in order to predict likelihood of breast cancer recurrence. As with pharmacogenomics, the appropriate evaluation of these tests is not yet resolved. Oncotype Dx has been proposed as a means to determine which women with early stage breast cancer require chemotherapy, based on large scale retrospective analyses. The need for prospective evaluation of testing outcomes is a matter of debate. This form of testing is

presumed not to involve genetics professionals; however, it is conceivable that future gene expression profiles could measure inherited rather than acquired variation, with broader genetic implications.

### Genetic susceptibility testing

Genetic susceptibility testing also raises questions regarding evaluation and appropriate use. When is genetic risk information useful and when is it harmful? Who decides? In light of the large volume of risk information that will flow from genomic research, these questions are critically important for health care policy. An important question is whether susceptibility tests should be viewed as helpful information, evaluated primarily for their predictive value, versus measures of risk that should be used only if they are proven to lead to interventions that improve health outcomes. Factor V Leiden, one of the few genetic susceptibility tests now in clinical use, illustrates the challenge: the test is widely ordered despite the fact that there is no evidence to suggest that FVL testing should routinely direct clinical management or will improve health outcome. This observation could be interpreted to mean that the test is being used inappropriately, or that it is providing information that clinicians and patients find of value. In addition, FVL testing will rarely identify individuals with very high inherited risk (e.g., FVL homozygotes), for whom specific therapy and referral to genetic counseling may be appropriate.

Consideration of genetic susceptibility testing leads to fundamental inquiries about the purpose of health care, incorporating how broadly “health” and “health outcomes” should be defined, and the limits that should be set on the use of health care resources. Genetic susceptibility testing also poses questions about delivery of care, including the degree to which current systems of primary care can be better focused on prevention. And to the extent that prevention can be made a central focus of primary care, rigorous questions will need to be asked about the value added by knowledge of genetic risk: e.g., if the health care system were already maximizing efforts to promote healthy diet, how would knowledge of genetic risk for diabetes or coronary artery disease assist patients or providers?

Storage and retrieval of genetic susceptibility information also pose challenges. The information will need to be readily accessible in all places where a patient receives health care if it is to be maximally effective. At the same time, appropriate privacy protection will be essential.

### **Provision of services**

As discussed above, genetics professionals provide a range of services to assist families and other clinicians to care for patients with genetic diseases (Table). Patient care for rare genetic diseases is likely to be best in centers with substantial experience treating these disorders. Often the specialty center is the only source of care within a state or large geographic area. The provision of genetic services relies on appropriate referral to genetic services, effective coordination of specialty services, and interdisciplinary management of patients with genetic disorders.

Since genetics professionals are not involved in primary care, they rely on other providers for patient referral. The provider seeing the patient for an initial complaint must recognize the

possibility of a genetic disease and know where to refer the patient. For this reason, primary care providers and local specialists need sufficient background knowledge of genetics, and appropriate access to point of service information, to make appropriate and timely referrals.

Particularly as advances are made in diagnosis and treatment of genetic disorders, specific referral to genetic services, from both the specialty center and primary care, is also important. Clinical geneticists are most likely to know if a diagnostic test is now available for a condition previously diagnosed on a clinical basis, or if new treatments or surveillance strategies are now available for a given condition. Clinical geneticists often play a key role in diagnosis and are increasingly playing a role in guiding and coordinating patient management, as discussed above. Furthermore, a specialist may be able to provide disease management after diagnosis but may be unprepared to counsel the family about mode of inheritance, genetic testing options for family members at risk or other services, such as prenatal diagnosis and other reproductive options.

Another issue is the need for genetics professionals with the expertise to provide services to adults with rare genetic disorders. This need stems from recently improved survival rates for many genetic disorders and the greater importance placed on long-term follow-up and management, as discussed above.

One of the biggest challenges in the delivery of genetic services is effective coordination of the different components of service for patients with genetic diseases and their families. This challenge encompasses communication between different specialties to ensure effective sharing of care among primary care provider, specialty provider, and clinical genetics. It often includes the challenge of coordinating across significant distances, and taking into account different funding mechanisms for the different components of care. This challenge is often poorly addressed in the current U.S. health care system.

To address this need for effective coordination of care for patients with genetic disorders, interdisciplinary clinics organized around a specific genetic diagnosis or family of diagnoses have been developed, like specialty clinics for cystic fibrosis, neuromuscular diseases, or PKU. Other interdisciplinary clinics are organized around broader diagnostic categories, some of which may have a genetic origin. Examples include hearing loss clinics and maxillo-facial clinics. Depending on the diagnosis, the geneticist may be asked to play a coordinating role. Progress in understanding the genetic contribution to common diseases is likely to lead to a new kind of referral clinic in which a multidisciplinary team, incorporating genetics professionals, provides care. The geneticist will likely be brought in as an expert or consultant to address specific issues about diagnosis, interpretation of genetic susceptibility test results, and counseling. Such clinics already exist in cancer genetics, for example.

### **Location of services**

Genetic services are usually provided in clinical genetics clinics based in academic medical centers or tertiary referral centers. One of the important reasons for locating clinical genetic services in academic medical centers is that they are poorly reimbursed. Often it is possible to deliver them effectively only because the salaries of the genetic counselors and clinical

geneticists are largely covered by research activities, with clinical care representing only a small fraction of their effort.

The increasing numbers of genetic tests now available will result in greater demands for the services of genetics professionals. Efficiencies may be hard to achieve in the absence of innovative approaches to support; for example, genetics professionals can often provide effective consultation to primary care providers and specialists via telephone and email, to allow them to complete the initial stages of work-up in a patient suspected of having a genetic disease. This consultation can limit inappropriate referrals and reduce time demands on genetics clinics, but is not usually reimbursed.

### **Barriers to integration of genetic services in practice**

The evolving role of the clinical geneticist must find a balance between the trend for clinical geneticists to be increasingly involved not only in diagnosis but also in offering guidance on patient management and letting other health professionals take on the diagnosis and treatment of some genetic disorders because the clinical genetics workforce is limited in numbers. Although some common genetic disorders are already mostly under the care of other health professionals (for example, cystic fibrosis), it will be difficult to decide, among some other disorders, which ones need to be diagnosed and/or managed by clinical geneticists and which ones can be taken on by other specialties. Clinical geneticists are not the first to struggle with what should be the boundaries of their practice (Greendale and Pyeritz, 2001). This dilemma has been compared to the one faced by infectious disease specialists in the last century. As new tests and antibiotic treatments became available, infectious disease specialists started to focus on the diagnosis of complex cases and less common disorders, as well as the use of the newest therapies, while the most common diseases and their treatments were dealt with by primary care providers and other specialists (Guttmacher, 2001). The advent of new technologies led to a redefinition of the specialists' role, not their disappearance.

For clinical geneticists to continue providing services, reimbursement issues must be resolved. Current reimbursement practices do not cover the costs associated with the provision of genetic services (Pletcher, 2002). The majority of clinical geneticists receive at least part of their income as a salary, and only 6% receive most of their income from traditional fee for service (Pletcher, 2002). In clinical genetics, patient evaluation, counseling, and education require more time than in other specialties, but reimbursement does not reflect this (Howell, 2002). Furthermore, counseling services provided by genetic counselors are often not reimbursed. Genetic counselors must be paid out of other sources of income. Academic clinical genetic laboratories used to be an important source of income for clinical genetics services, but competition from commercial laboratories has made them less profitable. Clinical genetic services cannot be maintained in the long run using the current reimbursement practices.

Because clinical genetics services are located in tertiary care centers, they rely entirely on referral from other providers for their practice. Primary care providers and other specialists need to be aware of the availability of genetic services in their area. They also need to be better educated about appropriate use of genetic services and indications for referral to a clinical

geneticist (Taylor, 2003). Clinical geneticists might need to come up with new ways to provide services to increase their visibility and availability; the use of phone or telemedicine consultations with providers and/or patients could reduce the number of unnecessary referrals while increasing the number of cases addressed by the clinical geneticist.

Because the number of clinical geneticists and other genetics health professionals is limited, some areas have little or no access to genetic services. Incentives must be in place to attract more qualified individuals into the field of clinical genetics and more genetic professionals to underserved areas. The use of telemedicine could also increase access to genetic services in underserved areas, but assumes that there are genetic professionals elsewhere who have the time and resources to offer telemedicine consultations. It must also take into consideration that not all services can be provided using telemedicine, and that a subset of patients will still have to travel to see the genetics specialist at a tertiary care center. In considering these challenges, however, the emerging uses of genetics testing and technology need to be considered. Some are readily integrated into existing health care while others may require specific efforts to define appropriate use and the role of genetics professionals.

### **Other innovative uses of genomic technology**

Genetic technology has also entered the health care system in a different way, as the source of improved health care tools for various health care problems, often based on genomics analysis of pathogens. The most obvious example is the use of DNA-based tests to identify microbial pathogens, providing more accurate and rapid means to determine the cause of many infectious diseases. Typically, the testing process itself is simple, requiring no specialized expertise.

Innovative genome-based therapeutics are also part of this trend. For example, fomiversen, an antisense oligonucleotide that binds to the messenger RNA of an essential protein of cytomegalovirus, can inhibit protein expression. It has proved to be an effective therapy for cytomegalovirus infections of the eye in AIDS patients. More innovations of this kind are likely; in general, they will be integrated into health care by the same route as non-genetic technical innovation, generally by direct comparison to the technology they are replacing, and will not necessarily change the structure or process of health care delivery.

### **Conclusions**

Although clinical geneticists still “specialize in genetic disorders and conditions,” there have been tremendous improvements in their knowledge about these disorders and the tools at their disposal to diagnose and treat them. As the role of the clinical geneticist evolves to integrate these advances into clinical services, their relationship with other providers will change.

Primary care providers and other specialists need to be aware of the availability of genetic services, learning how and who to refer for genetic evaluation. In addition, clinicians not trained in genetics will need to develop the skills to use certain genetic services independently – notably pharmacogenomics and genetic susceptibility testing.

Clinical geneticists have to develop new ways to increase access to their services in a context of limited resources, and also participate in the development of robust practice standards for the use of genetic tests and technologies in other clinical settings, in partnership with the appropriate medical specialties. Interdisciplinary collaborations will become more common. Reimbursement practices will have to change to reflect these new developments to sustain the provision of genetic services.

**Table: Roles of Genetics Professionals**

	Clinical roles	Usual site of practice
Clinical Geneticists	<ul style="list-style-type: none"> <li>• Diagnosis of genetic disease</li> <li>• Consultation regarding clinical management</li> <li>• (Rarely) primary care of patients with genetic disease</li> <li>• Supervision of genetic counselors (not required in all states)</li> </ul>	<ul style="list-style-type: none"> <li>• Academic medical centers</li> </ul>
Genetic counselors and nurse geneticists	<ul style="list-style-type: none"> <li>• Evaluation and counseling of patients related to risk for genetic disease in themselves or family members</li> <li>• Counseling before and after genetic testing</li> </ul>	<ul style="list-style-type: none"> <li>• Academic medical centers</li> <li>• Specialty referral clinics</li> <li>• State-supported genetics clinics</li> </ul>
Genetics laboratory professionals	<ul style="list-style-type: none"> <li>• Perform or supervise laboratory procedures for genetic tests</li> <li>• Interpret test results</li> </ul>	<ul style="list-style-type: none"> <li>• Academic medical centers</li> <li>• State laboratories</li> <li>• Commercial laboratories</li> </ul>

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